Clinical Practice Guideline for the Management of Patients with Autism Spectrum Disorders in Primary Care

NOTE:

More than 5 years have passed since the publication of this Clinical Practice Guideline, and **its validity has not been reviewed**.

Therefore, **it is unclear whether its recommendations remain valid**; they should therefore be treated with caution and applied in accordance with **clinical judgement and the individual circumstances of each patient**.

CLINICAL PRACTICE GUIDELINES IN THE SPANISH NATIONAL HEALTHCARE SYSTEM
MINISTRY OF HEALTH, SOCIAL POLICY, AND EQUALITY

MINISTERIO DE CIENCIA E INNOVACIÓN
MINISTERIO DE SANIDAD Y POLITICA SOCIAL
Plan de Calidad para el Sistema Nacional de Salud
SaludMadrid
Agencia Laín Entralgo para la Formación, Investigación y Estudios Sanitarios
Comunidad de Madrid

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CLINICAL PRACTICE GUIDELINES IN THE SPANISH NHS
MINISTRY OF HEALTH AND SOCIAL POLICIES

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This CPG is an aid to decision making in health care. It is not mandatory nor replaces the clinical criteria of health care professionals.

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Presentation

Care practice is increasingly complex due to several factors, among which is the increase of scientific information available. Science, something that is constantly changing, requires constant updated knowledge in order to meet the needs of health care and disease effectively and safely.

In 2003, the Spanish NHS Interterritorial Council created the GuíaSalud project whose final aim is the improvement of quality in clinical decision-making. For this reason, a register of Clinical Practice Guidelines (CPG) in the National Health System has been developed based on explicit criteria generated by its scientific committee.

In early 2006, the Directorate General of Quality Assurance Agency for Health developed the National Quality Plan for the National Health System, which includes 12 strategies. The purpose of this Plan is to increase the cohesion of the system and help ensure the highest quality healthcare to all citizens regardless of their place of residence. This plan includes the development of CPGs by different agencies and experts groups on prevalent pathologies related to health strategies.

In this framework, this guide has been created to treat patients with Autism Spectrum Disorders (ASD) in Primary Care.

Mental disorders are a major social and economic burden, both by its frequency, coexistence and co-morbidity, as well as by the disability they produce. In the case of ASD, there is a considerable impact on not only the development and welfare of those affected, but also their families. The chronic nature and severity of these disorders require the creation of a plan for early detection and personalised and permanent multidisciplinary treatment throughout the life cycle, with significant social and economic costs.

The evidence that ASDs are more common than previously thought has increased in recent decades. Knowledge of this fact supports the development of this Clinical Practice Guideline on Autism Spectrum Disorders for Primary Care, as health professionals in this area of care, in coordination with other professionals (education, social services), are key actors of early detection and subsequent establishment of the best comprehensive care for children. They are also best placed to provide information, support and guidance to the family.

We hope this guide will facilitate the work of professionals and improve the quality of care provided to children with ASD and their families.

PABLO RIVERO CORTE
Directorate General of the Quality Agency of the NHS

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Authorship and Collaboration

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Development Group of the CPG for the management of patients with Autism Spectrum Disorders in Primary Care

Abad Cristina Sanz. Nurse. Primary Health Care Team "José M ª Llanos" Primary Health Centre. Madrid
Josep Artigas Pallares. Paediatric neurologist / psychologist. Sabadell Hospital. Barcelona
Diana Milena Bohorquez Ballesteros. Psychologist. University of Salamanca. Scholar at the University Institute on Community Integration (INICO). Salamanca
Ricardo Canal Bedia. Psychologist. University of Salamanca. Professor of Behavioural Disorders. INICO. University of Salamanca
Ángel Díez Cuervo. Neurologist / Psychiatrist. Medical Adviser of APNA-FESPAU and JARES Associations. Madrid
M ª Inmaculada Dorado Siles. Mental Health Nurse. Mental Health Unit University Hospital "Virgen del Rocío". Sevilla
Cristina Espeso Sanz. Patient Representative. Spanish Federation of Associations of Parents of Autistic Children FESPAU. Madrid
José Galbe Sánchez-Ventura. Paediatrician. Primary Health Care Team "Torrero La Paz" Primary Health Centre. Zaragoza
Javier Gracia San Román. Physician in Preventive Medicine and Public Health. Health Technology Assessment Unit (UETS). Lain Entralgo Agency. Regional Ministry of Health. Madrid
Juana María Hernández Rodríguez. Psychologist. Specific Team on Severe Development Alterations
Ministry of Education of the Autonomous Community of Madrid
Raquel Luengo González. Nurse / Anthropologist. Health Technology Assessment Unit (UETS). Lain Entralgo Agency. Regional Ministry of Health. Madrid
Francisca Menchero Pinos. Paediatrician. Primary Health Care Team José M ª Llanos Primary Health Centre. Madrid
Ruth Vidriales Fernandez. Psychologist. Patient Representative. Spanish Autism Confederation-CAE. Madrid

Jose Vivancos Mora, neurologist, coordinator of the Stroke Unit, Hospital Universitario de La Princesa, Madrid. Cerebrovascular Disease Study Group, Spanish Society of Neurology (SEN)

Coordination

Francisco Javier Gracia San Roman and Raquel Luengo González. Health Technology Assessment Unit (UETS). Madrid

External review

Rosario Alonso Cuesta. Child Psychiatrist. Child Mental Health Unit.

University Hospital "Virgen del Rocío". Sevilla
Celso Arango López. Child and adolescent psychiatrist. Integral Health Care of Autism Spectrum Disorders (AMI-TEA). University Hospital "Gregorio Marañón". Madrid
Leticia Boada Muñoz. Psychologist. Integral Health Care of Autism Spectrum Disorders (AMI-TEA). University Hospital "Gregorio Marañón". Madrid
Mercedes Elizalde Usechi. Paediatrician. "Orcoyen" Primary Health Center. Navarra
Soledad Estébanez Ruiz. Paediatrician. "Los Cubos" Primary Health Center. Burgos
Emilio Fernández Alvarez. Paediatric neurologist. Hospital "Sant Joan de Déu". Barcelona
Maria Frontera Sancho. Psychologist and Educator. University of Zaragoza. Professor of the Faculty of Education. Zaragoza
Carlos Garcia Gil. Parent of a child with ASD. Madrid
Santiago García-Tornel Florensa. Paediatrician. Hospital Sant Joan de Déu. Barcelona
Francisco Gonzalez Ramos. General Practitioner. "Villares de la Reina" Primary Health Center. Salamanca
Manuel Ángel Herrera Martiache. General Practitioner. "Villares de la Reina" Primary Health Center. Salamanca
M ª Concepción Martín Arribas. Nurse. Research Institute on Rare Diseases. Carlos III Health Institute. Madrid
Mara Parellada Redondo. Child and adolescent psychiatrist. Comprehensive Health Care of Autism Spectrum Disorders (AMI-TEA). University Hospital Gregorio Marañón. Madrid
José Luis Peña Segura. Paediatric neurologist. Hospital Miguel Servet. Zaragoza
Pedro Manuel Ruiz Lázaro. Child and adolescent psychiatrist. Universitary Hospital "Lozano Blesa". Zaragoza
Nieves Ines Sanchez-Camacho Gomez-Cambronero. Social Worker. Primary Care Team of "José M ª Llanos" Primary Helath Centre. Madrid
Javier Tamarit Cuadrado. Psychologist. Quality Manager of the Confederation Spanish Organizations for Persons with Intellectual Disabilities Madrid

Collaborating scientific societies and institutions

This CPG has the support of the following organisations:

Spanish Society of Professionals in Autism - **AETAPI**
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Spanish Confederation of Organisations in favour of People with Intellectual Disabilities - **FEAPS**
Spanish Society for Paediatric Neurology - **SENP**
Spanish Federation of Community Nursing and Primary Care - **FAECAP**
Autism Confederation Spain - **CAE**
National Association of Mental Health Nursing - **ANESM**
Spanish Association of Child and Adolescent Psychiatry - **AEPNYA**
Spanish Society for Family and Community Medicine - **semFYC**

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Declaration of interest: All the members of the Development Group have made a declaration of interest presented in Appendix 11..

Questions to answer

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DEFINITION, CLINICAL MANIFESTATIONS AND CLASSIFICATIONS

- What is the definition of ASD?
- What is the aetiology of ASD?
- What is the co-morbidity of ASD?
- What are the clinical manifestations of ASD?
- What is the classification of ASD?

DIAGNOSTIC CRITERIA

- What are the diagnostic criteria of ASD?
- Are the diagnostic criteria established in the DSM-IV-TR or ICD-10 manuals on ASD diagnosis useful?

EARLY DETECTION

- Why is early detection important?
- How should the monitoring of a healthy child in Primary Care be carried out to detect early problems in the further development?
- Are there warning signs / specific criteria for suspicion of ASD useful for early detection in Primary Care?
- May the parent's warning be effective in Primary Care for risk assessment of ASDs?
- What rating scales of suspected ASD are most effective?
- Do we need a more comprehensive monitoring in risk groups?
- What is the minimum age of suspicion?

ASD SUSPICION MANAGEMENT STRATEGIES IN PRIMARY CARE

- What are the steps to follow if there is any suspicion of ASD? What are the referral criteria of ASD? - Algorithm

KEY ASPECTS TO CONSIDER WHEN PROVIDING INFORMATION TO PARENTS

- Does the information given to parents of children with suspected ASD amend their satisfaction and their ability to cope with the problem?
- What are the aspects to consider in relation to parents when providing information if there is a suspicion of ASD, and after a diagnosis of ASD?
- What is the basic information that Primary Care professionals are required to provide to parents of children with suspected or newly diagnosed ASD?

STRATEGIES FOR MONITORING CHILDREN WITH ASD IN PRIMARY CARE

- How to plan the visit of children with ASD in Primary Care?
- What issues should be explored / monitored in the Primary Care visit of children with ASD?

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Recommendations

Diagnostic Criteria

C	It is recommended that professionals involved in the detection of children with ASD, especially those with little experience, use the diagnostic DSM-IV-TR and / or ICD-10 manuals

Early detection

C	Early identification of children with ASD is recommended as part of the monitoring healthy child care process
D	Health and educational professionals should regularly discuss with parents the psychomotor, intellectual and behavioural development of their children (at least between 8-12 months, between 2-3 years and between 4-5 years old) as part of the monitoring of the healthy child
D	Health professionals should incorporate a high level of vigilance in the fields of social, play, language and behaviour development for the early identification of ASD and related disorders
√	In the monitoring of healthy child development in Primary Care, to help detect any developmental disorder, including ASD, it is recommended the use of scales (such as the Haizea Llevant scale developed in our context) (Appendix 3)

Warning signs

D ,√	Healthcare professionals should monitor the development of healthy children taking into account the "warning signs" proposed (Appendix 4): - Immediate warning signals - Compendium of ASD warning signs according to age periods

Warnings from parents

C	Concerns or warnings from parents regarding their child's development should be valued as much as the very presence of abnormal features
C	Take advantage of any visit (routine monitoring or due to illness) to consult the parents on concerns regarding their child's development
√	Professionals can use the items of the PEDS scale to direct questions about parental concerns (Appendix 5)

Rating scales of suspected ASD

C	Population screening for ASDs is not recommended with the instruments developed at thismoment, although the capability of the M-CHAT and the Autonomous Scale is being investigated

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B, C	We recommend the M-CHAT instrument as a useful tool to confirm clinical suspicion of ASD in children between 16 and 30 months (Appendix 6)
B, C	We recommend using the Autonomous Scale, in children older than 5 years as a useful tool to confirm clinical suspicion of Asperger's disorder (Appendix 7)

Monitoring in risk groups

C, D	It is recommended to take into account those factors associated with the development of ASD in regular visits: perinatal factors (consumption of drugs or teratogenic substances, perinatal infections, gestational diabetes, gestational bleeding, foetal distress, prematurity, low birth weight), metabolic diseases, older parents, parents with a history of schizophrenia or affective psychosis and a family history of ASD (siblings with ASD)
C	Professionals should carry out a close monitoring of the development of those siblings of children with ASD as these are a risk group

Minimum age of suspicion

C	Practitioners should monitor the presence of any warning signals from at least 6 months of age
√	Given that there is no evidence of a minimum age for alterations, referral to Specialised Care may be considered at any age when there is a suspicion of ASD

Aspects to consider when providing information to parents for suspected ASD

√	Practitioners should provide information to parents about what is appropriate child development
D	It is recommended to always address any concerns parents may have about the behaviour or development of their child to improve their satisfaction with the treatment received by the professional
Q	It should be remembered that new parents or parents without previous references may have more difficult to detect disorders in their children's development
√	When there is a suspicion of ASD, parents should be warned but not alarmed, making them aware of the importance of immediate evaluation
√	Professionals should provide correct information to help parents realize their child's problems, because some families have difficulty recognizing, understanding and accepting the disorder, especially when professionals are giving completely unexpected information
√	It is recommended NOT TO USE the terms "disorder" or "autism" (since a diagnosis has not been made yet), and use expressions such as: your child seems "not to have a communicative and social development appropriate for his or her age"
√	Action must be taken to the feelings of fear and denial of parents by being positive, non-judgmental and actively listening to concerns about the referral to Specialised Care

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D	If there is any suspicion of ASD, one must be agile and provide information on the diagnostic process (referral), to increase satisfaction and confidence of parents and reduce their uncertainty (Appendix 8)
√	The Health Care Team, especially the social worker, should take an "enabling" role when providing and interpreting information together with families

Aspects to consider when providing information to parents after a diagnosis of ASD

Q	Professionals must accept and understand the first emotions resulting from the diagnosis as part of the process of accepting the new reality
D	Professionals should provide objective and simple information to parents about what are ASDs, their aetiology, and respond to any questions that may apply, so that they can control the situation as soon as possible and increase their ability to cope with it (Appendix 9)
D	Professionals should be wary of burnout, depression and stress phenomena of the primary caregiver, usually the mother
Q	Professionals should be positive in the messages that are transmitted so that the process of adaptation and acceptance of families is effective, since they must rebuild their social conceptualization of what it means to have a child with autism, as well as acquire management skills
√	The Health Care Team, especially the social worker, should take an "enabling" role when providing and interpreting information together with families

Management and monitorization of children with ASD in Primary Care

√	Professionals should take account of the symptoms and behavioural disorders of children with ASD to prepare, in coordination with parents, the visits of the child to prevent overstimulation or confusion and potential health problems or risk behaviours
C	It would be useful to register children with ASD by maturation level (IQ or developmental level) as it is a key prognostic factor, which determines the global evolution (learning, communication, social skills)
B	Professionals can use visual support interventions such as the use of pictograms, cartoons, dolls imitation, to support communication processes in children with ASD. Example: to show them the instruments, procedures and techniques, exploration of pain and other symptoms
D	Professionals should use short simple sentences, without double meanings to communicate previously to the child what he or she will do, and use direct orders when asking them for something, using the support of parents to understand and communicate

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√	In the visit of children with ASD, Primary Care professionals should: – Coordinate with parents so that they anticipate the visit to the child – Try not to wait for a long time in the waiting room, especially if it is full of other patients – Trust in the management criteria of parents – Welcome the child in a calm and orderly environment – Reinforcement with prizes (toys, stickers and others) may facilitate future visits – In case a certain procedure that involves physical contact is required, it is necessary to assess risk / benefit of doing so. For example, blood tests, for which he or she should be as much prepared as possible, accepting more flexible rules such as allowing the parent to accompany the child or adapting the procedure if necessary – Be aware of disorders and behavioural sensitivity to detect: masked symptoms such as pain, certain harmful or dangerous habits like inhaling or ingesting toxic substances, self-harm – Adjust the format of the medication to the child's preferences as much as possible (i.e., syrup instead of tablets)
D	Professionals should monitor the nutritional status of children with ASD, recommending dietary supplements if necessary or even referring
D	Professionals should manage the gastrointestinal problems of children with ASD in the same way as those of children without ASD, taking into account that the existence of functional gastrointestinal problems (constipation, bowel disorders, and others) can have major adaptive and behavioural consequences than in population without ASD
√	Professionals should inform parents about healthy habits such as diet or exercise, leisure and free time activities, selecting those which, within the limits of the family and those provided by the community, provide enjoyment and child welfare
D	Monitor oral health of children with ASD. In case of eating toothpaste, do recommend one without fluoride. Patients and parents' associations can provide guidance on which dentists have experience attending children with autism, as sometimes certain changes in the procedures may be necessary. The same applies if ophthalmic check-ups are required
B	All children with ASD should follow the immunisation schedule established just like any other children, including vaccination of MMR (rubella, measles and mumps). Professionals should inform about the importance of this to parents
C	Professionals should monitor the presence of sleep disorders, using as a first step behavioural strategies to manage them and training parents on this issue
C	Professionals should inform parents that there is no evidence that ASDs are associated with digestive disorders
A, B	Professionals should inform parents that currently there is no evidence of the effectiveness of diets free of gluten and casein, secretin, vitamin B6 plus magnesium, Omega-3 fatty acid, dimethylglycine, hyperbaric oxygen therapy and music therapy for specific treatment of ASD

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C, D	Professionals should inform parents that currently there is no evidence of the effectiveness of antifungal therapy, chelating agents and immunotherapy
√	Professionals should inform parents that there are currently some "alternative treatments" that have no scientific basis to support their use and could be potentially dangerous, so they should be advised to consider only those interventions recommended by professionals

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1. Introduction

Mental health, as an indivisible part of health, contributes significantly to the quality of life and full social participation. Mental disorders are a major social and economic burden, both by their frequency, coexistence and comorbidity, as well as by the disability they produce. Mental illness is the second major cause of disease burden in societies with market economies, with no prospect of control of these figures, as a significant increase has been predicted[1]. In our context, it is confirmed as one of the main contributors to the loss of years of life free of disease, using as a measure of disease burden the rate of DALYs (Disability Adjusted Life Years)[2]. Therefore, mental health has required special attention by all those involved, reflected in the course of action of the document *"Mental Health Strategy for the National Health System"*[3]: health care to patients, coordination between institutions, scientific societies and associations, training of health personnel, promoting research and information and evaluation systems. The aim of this strategy is to improve the care of people with these disorders, reduce stigma and marginalisation to which they are subject and encourage prevention, early diagnosis, treatment, rehabilitation and social integration.

In the case of children with special needs *the Technical Report on health and social problems of children in Spain (2005)*[4] and the *White Paper on Early Care*[5], reiterated the priority of maximizing the development of those children who have deficiencies and encourage their academic and social integration and personal autonomy. In addition, it states that this goal requires adequately trained professionals working in interdisciplinary cooperation and coordination with parents and the institutional entities involved (health, educational, social).

Autistic Spectrum Disorders (ASDs) are part of mental health problems. ASDs are a number of neuropsychiatric disorders, classified as *"pervasive developmental disorders"* according to the diagnostic classification of DSM-IV-TR, which can be detected at an early age (by definition, autistic disorder is before 3 years old)[6]. This dysfunction has a major impact on not only the proper development and welfare of the individual concerned but also on the family, given the high burden of personalised care they need[7]. In addition, it must added, that the prevalence of such disorders has increased considerably, which together with its chronic nature and the severity of disorders, require a multidisciplinary customised and continuing treatment plan throughout the life cycle, under constant review and monitoring. This plan promotes the full potential of people with ASD and promotes their social integration and quality of life[8].

As with other mental health disorders or other disabilities, these are a problem that can cause stigma, which is further aggravated by a rare disorder to be socially visible, hidden or poorly understood by citizens and institutions as a whole. Families show feelings of isolation, invisibility and lack of knowledge of what are ASDs by the society in general[9, 10].

The diversity of clinical manifestations that can occur in ASD hinders early detection of symptoms consistent with such disorders[11], and consequently the diagnosis is delayed, which in many cases is not confirmed before the age of 3.

In addition, there is a lack of knowledge of the problem in medical and educational fields. In order to achieve a diagnosis as early as possible, it is very important to raise awareness of the importance of early detection of ASDs and provide tools to help an early detection and make decisions in this regard[12].

This document is the full version of *"Clinical Practice Guidelines for the Management of Patients with Autism Spectrum Disorder (ASD) in Primary Care (PC). Early detection and monitoring"*. The Clinical Practice Guideline (CPG) is structured by chapters that provide answers to the questions that have been stated at the beginning of this document. A summary of scientific

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evidence and recommendations is presented at the end of each chapter. Throughout the text, in the right margin the type of study and the possibility of bias in the literature reviewed are shown.

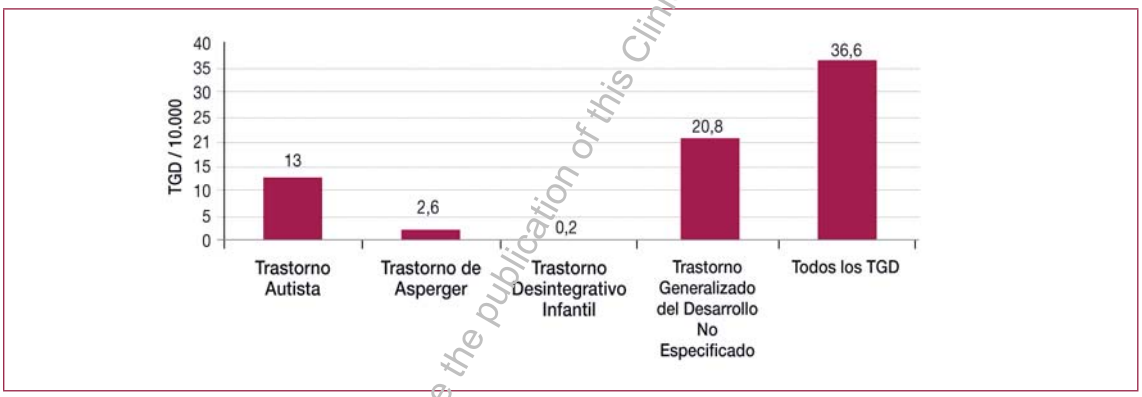
The material that presents the detailed information with the methodological process of the CPG (search strategy for each clinical question, guide tables) is available both on the website GuíaSalud as in the UETS, Health Technology Assessment Unit of Lain Entralgo Agency (Madrid). These websites also include the *Methodological Manual* on CPG *development process* which contain the general methodology employed[13]. Moreover, there is a condensed version of the CPG, to a lesser extent and with the appendices to the main CPG *full version*, a quick guide with the recommendations, scales and key algorithms, and a *version for parents and families of children with suspected and diagnosed with ASD*. These versions are found on the websites listed as the printed version.

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Prevalence of Autism Spectrum Disorders and burden of the disease

Autism has traditionally been considered a low prevalence disorder. The estimated figures from the 70s state around 4-5 per 10,000, but today the numbers have increased[14-17]. The estimated overall prevalence for autistic disorder, Asperger disorder, childhood disintegrative disorder and pervasive developmental disorder not otherwise specified are shown in Figure 1.

Figure 1. Prevalence of Pervasive Developmental Disorders– PDD / 10.000*



* Data from Fombonne15

More recent studies even reach a higher prevalence of 60 per 10000 for the entire autistic spectrum, as in the study in the population of New Jersey (United States), which provided prevalence data from a total of 67 cases per 10,000 children for the entire autistic spectrum, divided into 40 cases per 10,000 children for autistic disorder and 27 cases per 10,000 for Asperger's Disorder[18], or the study of Kent (United Kingdom) with a prevalence of 82.2 per 10,000 for the whole autism spectrum[19].

The increase in numbers has been gradual over the past 30 years. Some authors agree with the highest estimates and argue that it is due to a change in diagnostic criteria, now less restrictive to expand the spectrum (Asperger's Disorder was included in the manuals in the 90s), and an increase of the sensitivity of professionals to these disorders[20]. On the contrary, other authors suggest that current data does not fully support this hypothesis and the numbers should be monitored in the coming years to ensure that there really is no increase in the incidence of autism[15].

In the Spanish context, few studies have addressed this disorder in different Autonomous Communities. The results have not exceeded an estimated prevalence of 5 per 10,000 cases in Spain, but the data are from studies conducted between the years 1990 to 1998, there being no subsequent records[10, 21, 22]. One study calculated an estimate of the incidence in Spain by age group from the incidence rates of a studio in Australia, reaching an adjusted incidence rate of 8 per 10,000 for the group of autism and Asperger disorder in the 0-4 age group, and 3.5 per 10,000 in the age group 5-9 and 1.4 per 10,000 in the 10-14 years age group[23].

It has also been observed that men are more likely than women to suffer from this disorder, with an estimated ratio of 4:1, without significant differences in the proportion of cases of autism among the different social classes and cultures studied[24,25].

The onset age of these disorders is very early, in some cases the first signs have been observed during the first year of life, hence the importance of comprehensive monitoring of child development[26].

In regard to the effect of cognitive ability, according to data of some authors, intellectual disability among people with ASD was 29.8% (67% among people with autistic disorder, 12% among children with pervasive developmental disorder not otherwise specified and 0% among children with Asperger disorder)[17].

According to the survey by the INE (National Statistics Institute) on disabilities, impairments and health in the year 1999, in our country there are 3.8 million disabled people, of which some 4,500 people are diagnosed with autism (0.12 %)[27] . However, despite the chronic nature of these disorders, not all people diagnosed with autism are in a situation of dependency. Many children with ASD can achieve high levels of functionality and independence, depending not only on their characteristics or the severity of disorders (low or normal IQ, presence or lack of functional language), but also in terms of support received from an early stage, both to the child and the family.

The figures regarding the social cost of people with ASD are high. An economic review from 2007 estimated the total costs needed for an autistic person over his life at 3.2 billion dollars. This study is based on a review of studies from the United States, UK and Canada, by doing an age distribution according to direct costs (medical and nonmedical) and indirect (lost productivity in people with autism and the time parents devote to children with autism)[28]. Another study from 2006 compared the annual cost of medical services among children aged 2 to 18 years diagnosed with autism and without autism, and it estimated an annual cost is three times higher in the group of children with autism in comparison to those who do not suffer from this disorder[29]. The study was conducted with the population of a health plan in Northern California. They concluded that the use of health services and the cost of these were significantly higher for children with ASD compared with children without ASD[29].

According to a study on the overall ASD morbidity burden in Spain in 2003, 43,928 Disability Adjusted Life Years (DALY) were estimated, of which 33,797 were attributable to autistic disorder and 10,131 to Asperger disorder together with pervasive developmental disorder not otherwise specified. These figures were calculated on a reference population of 6,043,479 (children between 0 and 14 years in Spain in 2003, representing 14.5% of total population). A significant aspect of this study refers to the importance of early detection and early intervention of ASD. According to the same study, when the morbidity burden is adjusted after receiving a proper early intervention, experts estimate a reduction in the disability weight of 0.4 in the group with autistic disorder, and 0.25 in Asperger's disorder and pervasive developmental disorders not otherwise specified; representing a total reduction of 11,817 Disability Adjusted Life Years (26.95%). However, the real benefit of early intervention can not simply be assessed in terms of morbidity burden, as it also implies a reduction of family stress, increased coping ability of the problem and increased

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optimism in the future, so for this reason, the benefits are much higher than the simple reduction of Disability Adjusted Life Years[23].

Variability in clinical practice

Although ASDs are often seen in very early ages, not all professionals who evaluate healthy child development both in Primary Care (paediatricians, nurses, general practicioners), and the educational field (teachers and educators) have incorporated into their daily practice a surveillance system to detect developmental disorders effectively, so sometimes a first diagnosis may be delayed months or even years[30, 31].

Primary Care professionals may have little information or training to detect any developmental abnormalities, or these are not as present as other diseases or disorders of child development[32, 33]. Similarly, it is common not to know what instruments or scales are useful for early diagnosis of ASD. A report[30] on late diagnosis of ASD in Spain, involving 607 families from all over Spain, found that the family is the first to suspect that something was wrong in the development of their son or daughter when he or she is 1 year and 10 months and that the average age of first medical consultation is around 2 years and 2 months. 63% of paediatricians refer to specialised services before 6 months, but 21% do so after 6 months, and 16% never gets referring. This same study shows that although 72% of families said to have gone at some point to a public health visit (51% mention Primary Care) less than 38% have received a specific diagnosis in the public health care, as families end up turning to private care primarily arguing lack of knowledge about ASD in public health and private assistance being faster and better to confirm the diagnosis. This diagnostic delay is even greater in the Asperger disorder and autism without associated intellectual disabilities (sometimes called high functioning autism, although experts do not recommend the use of this term because it does not represent any diagnostic category), where symptoms can go unnoticed in the early years. In fact, the diagnosis of Asperger disorder, in children under 4 years can be very difficult. In Spain, data from a recent study[34] have shown that the diagnostic process usually takes several years and the age for diagnosis may be delayed up to 11 years; other international studies have shown the same delay for the group of children with ASD and in particular, with Asperger's disorder[35]. Therefore, it is necessary to encourage early detection taking into account the gradual range of symptoms and degrees of involvement of ASD, from the slightest alteration to the most serious developmental disorders.

Some professionals may think optimistically that the child has only mild signs of developmental delay that he or she is a bit "slow" but that will recover with time and reach the others[36, 37]. This favours delayed diagnosis, leading some parents to a situation of helplessness, visiting different medical consultations, to find a professional to guide the diagnosis[30]. The anxiety and uncertainty with which they live is very important during this period[38, 39].

Another study carried out in Spain in over 68 families and 109 health professionals describes the problems of accessibility for people with ASD to the health system[40]. 62.5% of families said they had difficulties in making the diagnosis of the disorder due to ignorance on the part of professionals (28.2%), the diversity of diagnoses (23.1%), delays in obtaining a diagnosis (15.4%), lack of knowledge about where to go (12.8%), little or no coordination between the professionals involved (12.8%), and the need to move to another city (7.7%). Concerning the time it took to get a definitive diagnosis, 41.4% of the families stated that it took more than four years to receive a definitive diagnosis. On the other hand, 80% of health professionals felt that they knew little or nothing about autism, and 20% knew some or very much about this disease.

Therefore, the priority is to provide the necessary information for early detection to both Primary Care practitioners (paediatricians, family physicians, nurses)[10, 41, 42], as well as educa-

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tional services, since these professionals are the ones who will be consulted first by parents if they detect any change, or they will be the ones to detect changes in the scheduled assessments of the child. Early detection will depend on these professionals having appropriate knowledge about the disorder[43,44].

It is also critical that parents obtain objective and reliable information, based on evidence, after a suspected ASD. Such information will reduce the stress and uncertainty in the process of referral to Specialised Care (SC), to both Primary Care professionals and parents, preparing them well for it.

Treating an ASD patient in Primary Care following diagnosis may be influenced by certain problems associated with these disorders and may require some consultation by the family to the paediatrician or Primary Care professional. It is therefore important to know the symptoms and co-morbidity associated with ASD, so that prevention and health promotion measures can be carried out, also facilitating the accessibility of children with ASD to health care[40].

Given the relevance of ASD, in some Autonomous Communities programs to provide specialised health care to patients with ASD are being created, thus eliminating all the barriers that the system has for these patients to be treated in accordance with their capabilities and features. One example is the AMI-TEA program (integral health care of patients with ASD), established in the Community of Madrid, University Hospital "Gregorio Marañón".

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The importance of early detection of autism spectrum disorders

Early detection will lead to early intervention in children with ASD and their families, which will help to reduce family stress, increase coping skills and the development of social adaptation of children in the future[45]. Institutions which support ASDs, professionals and relevant publications suggest that identification and early diagnosis are important (see Table 1)[46]:

Table 1: Benefits of early diagnosis of ASD[46]:

• prevents or rectifies erroneous diagnoses • reduces parents uncertainties • helps identify education options • helps in vocational guidance, and identifies services that facilitate this choice • provides access to resources, support and services • allows genetic counselling to families • allows adequate environmental support • facilitates contact with other people or relatives with ASD	• helps identify family support needs • helps people with ASD to understand themselves and others • helps others understand people with ASD, including family, couples and society • prevents future comorbidity problems • reduces isolation • guides the identification of the broader phenotype of ASD in family members

Early care resulting from early detection of health problems is justified by itself, since the therapeutic delay is always seen as a failure of the health system. Early care is defined as *"the set of interventions aimed at children of 0-6 years, the family and the environment, which aim to respond as soon as possible to the temporary or permanent needs presented by children with developmental disorders or who are at risk of suffering them. These interventions, which must consider the child as a whole, must be planned by a team of interdisciplinary and transdisciplinary orientation professionals"*[5]. Most of these teams are formed by psychologists, educational psychologists, speech therapists, physiotherapists, psychomotor therapists and neurologists. In the case of a developmental disorder, early intervention will at least reduce family stress and uncertainty about the future of the affected child.

This guide is intended, therefore, to be a useful tool for all professionals involved in early detection and monitoring of ASD in Primary Care, as well as parents and families involved in this care, so as to improve the quality of life of children with ASD and their families.

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2. Scope and objectives

The main purpose of this guide is to help health professionals in Primary Care to be able to perform an early detection and monitoring of children with ASD according to recommendations based on scientific evidence.

The topics covered in the guide are presented in the following specific objectives:

- Making Primary Care paediatricians and family physicians able to identify early developmental abnormalities and specific signs of suspected autism spectrum disorders in children, and to proceed with an appropriate referral for specialised care.
- Facilitate the Primary Care nursing staff and even other professionals related to early detection (teachers, counsellors) the ability to recognize early warning signs indicative of a developmental abnormality and specifically of an autism spectrum disorder.
- Provide primary health care professionals the right information to give to the family with a suspected case of ASD that requires referral and after the confirmed diagnosis.
- Train Primary Care professionals to act in a coordinated and effective way (each according to his professional duties) when monitoring children with ASD in Primary Care.

These objectives aim to improve the communication channels between Primary Care and Specialised Care to facilitate early diagnosis and expedite a customised care plan, hoping to improve the quality of life of the patients and their families.

The guide considers, within ASDs: autistic disorder, pervasive developmental disorder not otherwise specified, Asperger disorder, Rett disorder and childhood disintegrative disorder, taking into account that these are within the category of Pervasive Developmental Disorders (PDD) according to the classification of DSM-IV-TR diagnosis. The DSM-IV-TR considers ASDs as PDDs, so both names are equivalent. Currently, most authors do not consider Rett disorder and childhood disintegrative disorder within ASDs due to their special clinical features, however, given the scope of this guide, all PDDs will be considered within ASDs.

This guide focuses on children from 0 to 6 years old, a period that covers most of the diagnoses of ASDs[47] and which coincides with the Early Warning Surveillance period[5]. This timeframe excludes adolescents and adults.

Therefore, the guide is aimed primarily at professionals working in Primary Care (paediatricians, family physicians, nurses, social workers). Given the content of the document produced, the information will be useful for other professionals involved in the detection and treatment of ASDs, both within the educational field (teachers in general and Counselling Teams) and social services (Early Childhood Units).

It also targets parents who come to this first level of care. For the latter it is a tool that will allow them to know what are ASDs, the diagnostic process, mitigate the stress caused by the uncertainty, lack of information, and may manage reliable information so as to avoid guidelines, which have not been endorsed by evidence.

The resources currently available in Primary Care have been taken into account before finalizing the recommendations. This guide does not address issues related to the treatment of ASD. For more information about this issue, please consult the *Guide on good practice for the treatment of autism spectrum disorders*[48]. Given the diversity of clinical pictures, this guide does not make recommendations on issues that may be associated with ASD such as the treatment of epilepsy,

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hyperactivity, anxiety, depression, sleep disorder, eating disorders, aggression, self-harm, obsessive rituals, attention deficit disorder and others.

As this guide has a national approach, issues related to the organisation of health services in particular areas are not addressed, though it tries to establish some basic recommendations on criteria for referral to specialised care, so that the guide may also be disseminated to other professionals related to the care of ASDs.

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3. Methodology

The methodology used is set out in the *Manual for the Development of CPG* of the Ministry of Health[13].

The steps followed are:

- Constitution of the guide development group, composed of professionals from: Primary Care (paediatricians, family physicians and nurses), Specialised Care (neurologists, psychologists and psychiatrists for children and teenagers) and experts from the Health Technology Assessment Unit (UETS); besides two representatives of patients with ASD have participated within the development team group, from the early stages of the project, in the development of this guide.
- Constitution of a subgroup, with members of the guide's group for the development of the patient-oriented version.
- Definition of the scope and objectives of the guide.
- Formulation of clinical questions using the Patient / Intervention / Comparison / Outcome (PICO) format.
- Literature search in Medline, Embase, Pascal Biomed, Cochrane Plus, DARE, HTA, Clinical Evidence, INAHTA, NHS, PsycINFO, ERIC, CINDOC. Languages: English, Spanish. Population studier: children from 0 to 6 years old. Publication Year Limitation: limited to primary education. First we searched to find clinical practice guidelines (CPG) and then assessed their quality using the CPG assessment tool developed by international collaboration, Appraisal of Guidelines Research and Evaluation (AGREE). Two CPGs have been included as a secondary source of evidence to help answer some sections of the guide. In a second phase a search of systematic reviews, meta-analysis and evaluation reports in the databases mentioned above has been carried out. In a third phase, there has been an expanded search of primary studies (clinical trials, observational studies, studies on diagnostic and prognostic testing).
- Quality assessment of studies and summary of the evidence for each question following the recommendations of SIGN (Scottish Intercollegiate Guidelines Network) for intervention studies and NICE (National Institute for Clinical Excellence) for diagnostic studies.
- Formulation of recommendations based on "formal assessment" or "reasoned opinion" from SIGN and NICE (Annex 1). The controversial recommendations or lack of evidence were resolved by consensus of the development group.
- An external review of the guide was performed by a group of professionals selected for their knowledge of the pathology addressed, the methodology in the development of guides and the Primary Care setting.
- Various scientific societies involved were contacted: Asociación Española de Profesionales del Autismo (AETAPI) (Spanish Society of Professionals in Autism), Asociación Española de Pediatría en Atención Primaria (AEPAP) (Spanish Association of Primary Care Paediatrics), Federación de Asociaciones de Enfermería Comunitaria y de Atención Primaria (FAECAP) (Spanish Federation of Community Nursing and Primary Care), Sociedad Española de Neurología Pediátrica (SENP) (Spanish Society

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for Paediatric Neurology), Asociación Nacional de Enfermería de Salud Mental (ANESM) (National Association of Mental Health Nursing), Confederación Española de Organizaciones a favor de las Personas con Discapacidad Intelectual (FEAPS) (Spanish Confederation of Organisations in favour of People with Intellectual Disabilities), and the Asociación Española de Psiquiatría del Niño y el Adolescente (AEPNYA) (Spanish Association of Child and Adolescent Psychiatry). For the participation of patients, the associations involved are: Confederación de Autismo de España (CAE) (Spanish Autism Confederation) and the Federación Española de Asociaciones de Padres/Tutores de Personas con Autismo (FESPAU) (Spanish Federation of Associations of Parents/Guardians of Children with Autistic Disorders). All societies are represented by one of the development group members or external reviewers.

- In www.guiasalud.es the material that presents in detail the information with the methodological process of the CPG (search strategy for each clinical question, guides' tables) is available.

Update of the GUIDE

The UETS, responsible for the publication of the Guide will also be responsible for its update within a period of 3-5 years, or earlier, depending on the new evidence. This update is done through the incorporation of updated literature searches, particularly focusing on those aspects where the recommendations could be substantially modified. The methodology to carry out this update process is reflected in the *Methodological Manual Update for Clinical Practice Guidelines*, available at www.guiasalud.es

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4. Definition, clinical manifestations and classifications

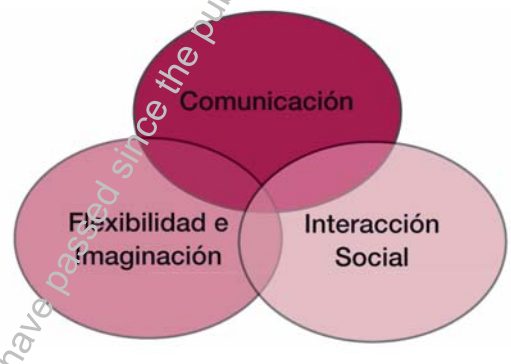
The questions that are going to be answered in this chapter are:

- What is the definition of ASDs?
- What is the aetiology of ASDs?
- What is the co-morbidity of ASDs?
- What are the clinical manifestations of ASDs?
- What is the classification of ASDs?

4.1. Definition

ASD is defined as a chronic neurological disorder with a strong genetic basis which is manifested from an early age in a series of symptoms based on a triad of disorders (Wing triad[49]) (see Figure 2) in social interaction, communication and lack of flexibility in thinking and behaviours (described in the DSM-IV-TR manual)[6]. The level of severity, type and age of onset of each of the criteria will vary from one individual to another, defining each of the diagnostic categories. Despite the ratings, no person suffering an ASD is similar to another one in terms of observable characteristics.

Figure1. Wing triad[49]



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4.2. Aetiology

Autism is a complex brain disorder that affects coordination, synchronisation and integration between different brain areas. The essential abnormalities of these disorders (social interactions, repetitive communication and behaviours and restrictive interests) are justified by multiple brain abnormalities, functional and / or structural, which are not always the same[50]. This approach provides the growing interest for the concept of autism as a spectrum of disorders that can cover different behavioural phenotypes, with varying degrees of intensity among the people who present it as well as during its evolution, which depend on the different brain areas involved with the possibility of being etiologically distinct.

This complexity of clinical manifestations suggests the existence of multiple causes. Recent advances indicate the importance of genetic factors and some possible environmental factors that lead to very early brain abnormalities.

The most widely accepted hypothesis is that the essential conditions of ASD arise during pregnancy (suggested before the 6th month), so they are often already present at birth, and manifest when they interfere with the normal course of development.

ASDs, therefore, have a primary etiological heterogeneity which is not always the same in all the families and individuals affected, and which can be influenced by environmental factors, especially in the early months of pregnancy, leading to nuclear disorder with different amplitude and severity, depending on the factors (genetic and environmental) involved[51-54].

Genetic factors

It is accepted that "autism is the most genetic disorder within the neuropsychiatric syndromes" and that it is primarily the result of the alteration of an interdependent set of genes, distributed in different parts of the genome, requiring the participation of a minimum number of genes, although they do not always coincide, for its development and manifestation[55].

The results of whole genome scans support the hypothesis that an individual must inherit at least 15 to 20 genes (genetic heterogeneity), which interact in a synergistic manner to express the complete phenotype of autism[56]. Of these genes, some act in all cases and others in various combinations that would influence in family variations and in the severity or the expression of the phenotype. The hypothesis proposed is that each of the genes involved provides a small amount of risk for the disorder and that only when that amount exceeds a certain threshold, the person has the full phenotype[57]. Other genetic aspects which contribute to autism are quantitative, polygenic and pleiotropic[58, 59]. All of these genetic features, as well as increasing comorbidity, explain the extensive phenotypic spectrum used to show these disorders. In addition, the balance between unfavourable quantitative traits and protective quantitative traits contributes to understanding the great variability among members of the same family for any of the traits of autism. The situation is even more complicated, taking into account that the expression of quantitative traits, both positive and negative, has a strong environmental influence.

The recurrence rate in siblings of people with autism is 2.2%, which can reach up to 8% when including all ASDs[17, 60-64], which means about 50-75 times the risk of the general population[65, 66]. Systematic studies performed on twins conclude that monozygotic twins (MZ) have a concordance rate (probability that both brothers have autism) above 60% for the full syndrome[67-71]. In addition, it has been found that MZ that are not consistent with autism, have cognitive and language disorders and / or social anomalies in a very high level, which can reach up to 76% contrasting with 10% in dizygotic[70,72]. These observations support the conclusion that autism is a polygenic disorder in which interactions between multiple genes can lead to the characteristic phenotype of

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ASDs in levels of variable intensity[65, 73].

Karyotype studies have shown that almost all chromosomes are involved in a 5-9% of people with autism[60,74-78]. In a person with autism, what is being specifically explored is the presence of "fragile X chromosome."

In addition to the karyotype, there are new genetic technologies (molecular genetics, ligament studies, polymorphism, fluorescent in situ hybridisation (FISH), microarrays comparative genomic hybridisation complete genome (CGH-arrays) and, more recently, multiplex ligation-dependent probe amplification (MLPA), used as complementary tests in the diagnostic process to detect associated syndromes / disorders.

There have been numerous studies in multiple-family groups and whole-genome scans[56, 79 - 89] that have suggested different regions of nearly all chromosomes, where loci and genes implicated in the aetiology of autism can be found.

In 5-8% of people with autism, monogenic disorders with specific phenotype and genotype characteristics, associated with a biological disorder that allow their individualisation are documented[51,52,54,90,91].These include (Table 2)[92-114]:

Table 2: Genetic disorders and autism

• Aarskog syndrome	• Lange syndrome
• Angelman syndrome	• Lesch-Nyhan syndrome
• Apert syndrome	• Lujan-Fryns syndrome
• Cohen syndrome	• Moebius syndrome
• Cowden syndrome	• neurofibromatosis type 1
• Tuberous sclerosis	• Noonan syndrome
• Cornelia de Lange syndrome	• Prader-Willi syndrome
• velocardiofacial syndrome	• Rett syndrome
• phenylketonuria	• Rubinstein-Taybi syndrome
• Steinert disease	• syndrome Smith-Lemli-Opitz
• progressive muscular dystrophy	• Smith-Magenis syndrome
• Duchenne disease	• Timothy syndrome
• Headd syndrome	• Sotos syndrome
• Hypomelanosis of Ito	• Williams syndrome
• Joubert syndrome	• Fragile X syndrome

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Epilepsy and autism

Epilepsy is a disorder associated with a high percentage of people with autism, with two major peaks of incidence, before the age of five and after puberty or adolescence, depending on various

factors (organic brain disorder, developmental regression, association of severe intellectual disability, certain genetic abnormalities...) that can cause a wider scope of neuropathology associated with epilepsy and autism, and in most cases, also associated with intellectual disability.

The EEG test is not indicated as a routine examination in children with primary autism without a history or clinical suspicion of seizures, because it does not provide diagnostic, prognostic or therapeutic data. However, it can be helpful, if there is history or there is a suspicion of epileptic seizures, and / or need to discard or characterise suspicion of specific syndromes.

Of all the pictures of epilepsy and epileptic syndromes[115], the one which is more interesting is the West Syndrome (WS). A significant number of children with West syndrome (up to 16%)[116] develop autism and other high percentage of children with autism (12-15%)[117] suffer epileptic seizures in early childhood, especially WS, without being clear whether WS or the aetiology of this (tuberous sclerosis complex, neurofibromatosis, phenylketonuria, among others) causes autism, being the percentage of children with West Syndrome and bitemporal involvement (71%) that evolves with autism even higher[53].

The Landau-Kleffner syndrome, acquired aphasia with epilepsy, is a neurological disorder which can only be diagnosed when the child has two core symptoms (100% of cases): an acquired aphasia that initially corresponds to a verbal auditory agnosia and specific disturbances in EEG. 75% of children undergo epileptic seizures between the age of 5 and 7, which disappear by the age of 10 to 15, and excited and disorganised behaviour but without the pathognomonic features of autism. The WHO in the ICD-10 classification (1992) points out that acquired aphasia with epilepsy and autistic disorder are two clinical disorders with excluding diagnostic criteria[118,119].

Neurochemical changes

Given the increasing acceptance that ASD may be enabled by neurochemical abnormalities that alter or modify the maturation of the CNS in some stage of early development, several possible neurochemical correlates of serotonin, oxytocin, dopamine, norepinephrine and acetylcholine are considered[120, 121].

Altered immune function

The potential role of maternal autoantibodies in the aetiology of some cases of autism has already been proposed. Recent studies indicate that the presence of IgG antibodies in maternal plasma during pregnancy against foetal brain proteins, together with genetic lability, may lead to global neurodevelopmental regression and thus may cause the development of early-onset autism[122-124].

Environmental factors

As described, although genetic factors are a priority in the aetiology of ASDs, these should not be the only causal factors, suggesting that there must be environmental factors, which act on the genotype, giving rise to the characteristic phenotype of ASD. The specific response of an individual to their environment depends on its genome and the genome's expression of a person is affected by the environment, i.e., the genetic and environmental factors are in a situation of reciprocity, not opposition.

Obstetric complications. Most obstetric complications are generally a consequence of foetal anomalies acquired in the early stages of embryonic development, rather than the cause of autism, as happens in a high percentage of difficult births of children without autism[125]. However, this environmental factor, although not being the underlying cause of autism may be a global aggravat-

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ing factor by potential associated disorders (intellectual disability, epileptic seizures, behavioural disorders, and others) due to genetic interaction.

Infectious diseases. In general, there is no evidence of inflammation or histological responses to infection, or demyelination in the CNS of individuals with ASD, consecutive to infection.

Triple-viral vaccine (measles, mumps, rubella). Numerous epidemiological studies have not proven a causal relationship between the triple-viral vaccine, "colitis autism" and ASDs, and do not support the proposed hypothesis of an association between the vaccine and ASD, according to researchers at Cochrane Vaccines Field that, after a review of 139 studies of the trivalent vaccine, concluded that no credible evidence was found about a relationship between the MMR vaccine and autism[126].

Exogenous brain opiates (gluten and casein). The inconsistency of the theories that say that children with autism metabolize incompletely the metabolic products of gluten and milk has been shown, so it does not justify the use of free gluten and casein diets[127].

Exposure to mercury (Hg). In relation to the theory of the triple-virus vaccine, it was believed that Hg, which was a preservative, was a causal factor in autism. Numerous studies have failed to confirm this conclusion. It is important to highlight that the elimination of thiomersal (ethylmercury) in vaccines has not provided a decrease in the prevalence of autism.

Toxic diseases. Intra-utero exposure to various toxic agents that may alter neurodevelopment and generate autistic-like foetopathy has been analysed. These include valproic acid and other antiepileptic drugs, cocaine, alcohol, thalidomide (although it is a drug which has been contraindicated in pregnant women for decades), lead, chronic exposure of the mother and foetus to low levels of carbon monoxide carbon and others, not always obtaining the same conclusions.

Misc. Theories have also been raised without any scientific basis such as and industrial and chemical substances present in the environment, which have entered the food chain, the use of pet shampoos by pregnant mothers, the chemical effects of rain, the exposure to TV, household chemicals, the lack of vitamin D for low sun exposure, being born in spring, parents who are scientists, engineers, physicists and mathematicians, candidiasis, the Borna disease, faecal clostridia, metallothionein dysfunction, the inability to metabolize phenolic amines, endocrine factors (increased estrogen, progesterone, testosterone in amniotic fluid).

4.3. Comorbidity and syndromic autism

In general, when talking about autism without further specification, it refers to idiopathic or primary autism without associated syndromes. In all cases of ASD, the diagnosis is made through direct observation of changes in the behaviour because there is no specific biological marker for autism. The additional evidence required are determined by the search for a specific aetiology or syndrome associated, resulting in a carrier of a "double syndrome", i.e. these children possess two diagnoses, one for ASD and another associated pathology[51]. We speak therefore of "syndromic autism" if the person suffering from ASD also has a related syndrome. When a disease or syndrome related to autism is presented, it does not necessarily mean that there is an etiological relationship between the two problems; therefore, it can only be considered a secondary autism case if a causal link between both disorders can be determined.

Children with ASD may have associated conditions such as: various metabolopathies, poisoning, infections, epilepsy[115,128,129] (the highest prevalence compared to the general population and much higher in ASD cases with cognitive disabilities[130]), motor problems, ocular and hearing disorders, hyperactivity, insomnia, and others[131, 132]. The associations of other mental health

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disorders such as obsessive-compulsive disorder, anxiety, depression and other mood disorders, especially in patients with Asperger disorder or autism without associated intellectual disabilities, which can start to manifest in adolescence have also been observed[131,133-135].

4.4. Clinical Manifestations

In all clinical cases, clinical manifestations occur in various aspects of the following areas: social interaction, communication and restricted repertoire of interests and behaviours. The manifestations of the disorder usually become apparent in the first years of life and will vary depending on the development and chronological age of the children.

The following table presents the major clinical manifestations that define ASD, including those listed in diagnostic manuals[6] and others not clearly stated in the manuals[136]:

Table 3: Clinical manifestations included in the manuals[6]

Impaired social interaction	Infants: eye contact, limited expressions and gestures. Resulting sometimes in a kind of selective deafness, as they do not usually react when called by their name. Young children: lack of interest in being helped, inability to initiate or participate in games with other children or adults, play games alone, inappropriate response in formal social relations.
Impaired Verbal and non-verbal communication	They do not use verbal and / or body as functional communication, even silence can exist in more severe cases. Some children begin language development in the first year of life but may regress after the second year and lose it. On the contrary, others suffer widespread delays in all aspects of language and communication (do not compensate by using gestures or mime). When language is present, there is a significant impairment in the ability to initiate or sustain a conversation: echolalia, confusion of personal pronouns (they refer to themselves in second or third person), verbal repetition of phrases or on a particular theme and abnormalities in prosody. They use a stereotyped and repetitive language.
Restriction of interests and stereotyped and repetitive behaviours	Absorbent concerns by one or more stereotyped and restricted patterns of interest that are abnormal in intensity or in its content. It is common: exaggerated emphasis on one activity, specific routines or rituals. They have little tolerance to changes in these routines. Usually acquire stereotyped and repetitive motor mannerisms such as shaking or twisting the hands or fingers, or complex movements of the body.
Other common clinical manifestations and not clearly set out in diagnostic manuals are136:	

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Unusual response to stimuli	Selective perception of certain sounds that translates into a lack of response to human voices or his or her own name and instead are extremely sensitive to certain sounds, even those considered mild for the majority of the people. The same occurs with certain visual, tactile, smell or taste stimuli. This sometimes results in eating disorders for choosing or rejecting certain flavours or textures, or rejection of certain garments by colour and feeling. Sometimes there is a serious dysfunction of pain sensation, which can mask medical problems such as fractures.
Behavioural disorders	Often they have difficulty focusing attention on specific subjects or activities, which they have not chosen themselves. Some children are often considered hyperactive or suffering from attention deficit problems, even reaching to significant levels of anxiety. Others respond to changes or frustrations with aggression or self-aggression.
Special Skills	Some children may show special skills in specific sensory, memory or calculation areas and in others that can make them stand out on fields such as music, art or mathematics.

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4.5. Classification of autism spectrum disorders

Today the terminology of ASD, although no qualifying term exists within the DSM-IV or ICD-10 manuals, it refers to Pervasive Developmental Disorders (PDD), being the first the one most used due to the dimensional flexibility that the ASD concept offers. The concept of ASD can be understood as a gradual range of symptoms, a "continuum" where one end is completely normal development and the other, more severe pervasive developmental disorders. This term was coined by Lorna Wing and Judith Gould in 1979 and represents a pragmatic rather than theoretical progress to the extent that it involves a detailed assessment of all the abilities and deficits of the subjects, thus helping to define their needs to support and implement the individualised treatment plan.

Basically, the two classifications (ICD-10 and DSM-IV) comprise the same diagnostic criteria; however, there are several arguments in favour of using the term ASD rather than PDD. The term "widespread" can be confusing or ambiguous, since it would imply involvement in all the aspects of development, which is not correct, since some people with autism have a normal development in some areas and affected in others.

The criteria included within the classification of DSM-IV are less specific than those from ICD-10. Among the disorders defined by DSM-IV, autism is the one, which is most defined, while Asperger's Disorder or Unspecified Pervasive Developmental Disorder are the least. Hence, false positives arise within these categories, which influence the level of research and acquisition of social and educational services[137]. In addition, the diagnostic categories offered by the manuals continue to be inadequate and difficult to use for the differential diagnosis of many "borderline" clinical cases.

The development group basically recommends the use of ICD-10 (WHO) and / or DSM-IV-TR (American Psychiatric Association) classifications for these disorders[6,138,139]. Below is a table (Table 4) comparing these diagnostic classifications:

Table 4: Comparison of diagnostic ICD-10 and DSM-IV-TR classifications

ICD-10	DSM-IV-TR / DSM-IV-TR-AP
Infantile autism (F84.0)	Autistic Disorder
Rett syndrome (F84.2)	Rett's Disorder
Other childhood disintegrative disorder (F84.3)	Childhood disintegrative disorder
Asperger syndrome (F84.5)	Asperger Disorder
Atypical autism (F84.1)	Pervasive developmental disorder not otherwise specified (including atypical autism)
Other pervasive developmental disorders (F84.8)	
Unspecified pervasive developmental disorder (F84.9)	
Hyperactivity disorder with intellectual disability and stereotyped movements	—

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Following **the diagnostic classification of DSM-IV-TR**, five types of disorders in PDDs are identified:

1. *Autistic disorder, infantile autism or Kanner's syndrome*. Their characterisation coincides with that described by Leo Kanner in 1943[140], where he describes manifestations in greater or lesser degree of the three main areas described above.

2. *Asperger's Disorder or Asperger Syndrome*. It is characterised by an inability to establish social relationships appropriate to their developmental age, with mental and behavioural rigidity. It differs from autistic disorder because it presents an apparently normal language development and without the existence of intellectual disability.

3. *Rett disorder or Rett Syndrome*. It differs from the previous ones in that it only occurs in girls and involves a rapid motor and behaviour regression* before the age of 4 (with stereotypic features such as "washing one's hands"). It is a disorder that appears in low frequency with respect to the foregoing. This disorder is associated with a severe intellectual disability. It is caused by mutations in the MECP2 gene[141].

4. *Childhood disintegrative disorder or Heller Syndrome*. It is a very rare disorder in which after a normal initial development, after two years and before the age of 10, a loss of pre-

* Note about specific regressions: in between 15 and 30% of children affected by ASD there is a period of stasis of development and even loss of acquired skills. The common moment when changes start to be noticed is between 15 and 19 months. Regression after normal development occurs between 2 and 4 years of age, being extremely distinctive in Rett syndrome and in infantile disintegrative disorder[142].

viously acquired skills is triggered. The most characteristic feature is the disappearance of the skills acquired in almost all areas. It is often associated with severe intellectual disability and an increase in EEG abnormalities and seizure disorders. It is suspected therefore that it is the result of an unidentified injury within the central nervous system.

5. *Pervasive developmental disorder not otherwise specified*. It includes all the cases, which are clearly not within the previous clinical pictures, or are incomplete or present inappropriate symptoms of autism as regards the onset age or existence of subliminal symptoms.

In the discussion on the classification of mental disorders in the future, the development group of the DSM-V is contemplating, among other things, the elimination of the Rett syndrome from the Pervasive Development Disorder chapter, and the introduction of a dimensional aspect to the categorisation of mental diseases. The latter aspect will be common to many other mental disorders and will not only affect autism spectrum disorders.

The following table presents a comparison of other medical and nursing diagnostic classifications that are being used for the ASD in our field.

Table 5: Comparison of other diagnostic classifications

ICD-9-CM	CIAP-2 (WONCA)	NANDA	DC: 0-3R
(299.0) Infantile autism	OTHER PSYCHOLOGICAL / MENTAL PROBLEMS (P99)	(111) Changes in growth and development (112) Risk of developmental abnormality	(710) Multisystemic Developmental Disorder
(299.8) Other unspecified psychosis from early childhood			
(299.1) Disintegrative psychosis			
(299.8) Other specified psychosis from early childhood			

- **ICD-9-CM (International Classification of Diseases):** Currently there is a newer version ICD-10, but in Spanish context, especially in the area of specialised care, version 9 is still being used, and it is included in its information system. It is available in electronic form on the Website of the Ministry of Health: http://www.msc.es/ecie9mc-2008/html/index.htm

- **CIAP-2 (WONCA)** - International Classification of Primary Care within the World Organisation of General Practitioners/Family Doctors, published in 1999, the Spanish version of the second edition (ICPC-2)[143]. This new version can sort by "episodes of care," a concept which is wider than the hospital concept of "episode of disease", as it can include: consultation reason expressed by the patient, health problems identified by professionals and care interventions or process.

- **NANDA** - International Classification of the North American Nursing Diagnosis Association[144, 145]. NANDA is a nursing scientific society whose aim is to standardise the nursing diagnosis by developing the nomenclature, criteria and taxonomy of such diagnoses. In 2002, NANDA became NANDA International. In the table, the defining characteristics and factors related to

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children with ASD, according to the criteria from nurses are presented. However, currently it is working with other NANDA diagnoses targeted to parents of children with ASD, which may also be helpful (see Annex 2).

- **DC: 0-3R MANUAL** - Diagnostic Classification on mental health and developmental disorders in early childhood from 0 to 3 years of age. There is no Spanish version of it, but in our context, it is used at the conceptual level in some Primary Care teams. Its creation by: Zero to Three / National Centre for Clinical Infant Programs in the United States is relatively new and it is hardly standardised, especially in our context. Studies about have concluded that it does have limitations regarding its use when addressing the criteria for different types of disorders and comorbidity diagnoses, as not being very accurate[146].

Finally there is also a diagnostic classification, little used in our context, which was developed for the detection of childhood mental disorders by the American Academy of Paediatrics: "The Classification of Child and Adolescent Mental Diagnoses in Primary Care: Diagnostic and Statistical Manual for Primary Care"[147]. This manual is intended for Primary Care professionals, but there are few studies examining its reliability and usefulness compared to other manuals.

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5. Diagnostic Criteria

The questions that are going to be answered in this chapter are:

- What are the diagnostic criteria of ASD?
- Are the diagnostic criteria laid down in the DSM-IV-TR or ICD-10 manuals useful for the diagnosis of ASD?

The diagnostic manuals are used at both Primary Care and Specialised Care, although it is necessary to comment that there is limited evidence on their use to improve the diagnostic process of children with ASD.

There are studies referring to the use of diagnostic criteria according to DSM-IV-TR and ICD-10; these showed that using the DSM-IV-TR and ICD-10 diagnosis the reliability of the diagnostic process increased, and the effects were greater if an inexperienced professional was involved in making the diagnoses. They also noted that the current criteria for Asperger disorder and autism have little discriminatory validity[148-151].

Here, we present the **diagnostic criteria** to be met by autism spectrum disorders to be considered as such by the **DSM-IV-TR**. The numerical codes next to each diagnostic category correspond to **ICD-10** and **ICD-9** consecutively[6].

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Diagnostic Criteria of F84.0 Autistic Disorder [299.00]

A. total of six (or more) items from (1), (2) and (3), with at least two of (1), and one of (2) and (3):

(1) qualitative impairment in social interaction, as manifested by at least two of the following characteristics:

(a) marked impairment in the use of multiple nonverbal behaviours such as eye-to-eye gaze, facial expression, body postures and gestures to regulate social interaction

(b) failure to develop peer relationships appropriate to developmental level

(c) lack of spontaneous seeking to share enjoyment, interests and objectives with others (i.e., not showing, bringing, or pointing out objects of interest)

(d) lack of social or emotional reciprocity

(2) qualitative impairments in communication as manifested by at least one of the following characteristics:

(a) delay in, or total lack of, development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gestures or mime)

(b) in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others

(c) stereotyped and repetitive use of language or idiosyncratic language

(d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

(3) restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:

(a) encompassing preoccupation with one or more stereotyped patterns of interest that is abnormal either in intensity or focus.

(b) apparent inflexible adherence to specific, non-functional routines or rituals,.

(c) stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements).

(d) persistent preoccupation with parts of objects.

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age of 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

C. The disturbance is not better accounted for by Rett Disorder or a Childhood Disintegrative Disorder.

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Criteria for the diagnosis of F84.2 Rett's Disorder [299.80]

A. All of the following characteristics:

(1) apparently normal prenatal and perinatal development

(2) apparently normal psychomotor development through the first five months after birth

(3) normal head circumference at birth

B. Onset of all of the following after the period of normal development:

(1) deceleration of head growth between the 5 and 48 months of age

(2) loss of previously acquired purposeful hand skills between the 5 and 30 months of age, with the subsequent development of stereotyped hand movements (i.e., hand-wringing or hand washing)

(3) loss of social engagement early in the course (although often social interaction develops later)

(4) poor coordination of gait or trunk movements

(5) severely impaired expressive and receptive language development with severe psychomotor retardation.

Criteria for the diagnosis of F84.3 Childhood disintegrative disorder [299.10]

A. Apparently normal development for at least the first 2 years after birth as manifested by the presence of age-appropriate verbal and nonverbal communication, social relationships, play, and adaptive behavior.

B. Clinically significant loss of previously acquired skills (before the age 10) in at least two of the following areas:

(1) expressive or receptive language

(2) social skills or adaptive behaviour

(3) bowel or bladder control

(4) play

(5) motor skills

C. Abnormalities of functioning in at least two of the following areas:

(1) qualitative impairment in social interaction (i.e., impaired nonverbal behaviours, failure to develop peer relationships, lack of social or emotional reciprocity)

(2) qualitative impairments in communication (i.e., delay or lack of spoken language, inability to initiate or sustain a conversation, stereotyped and repetitive use of language, lack of varied make-believe play)

(3) restricted, repetitive and stereotyped patterns of behaviour, interests and activities, which include motor stereotypies and mannerisms

D. The disturbance is not better accounted for by another Pervasive Developmental Disorder or Schizophrenia.

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Criteria for the diagnosis of F84.5 Asperger disorder [299.80]

A. Qualitative impairment in social interaction, as manifested by at least two of the following characteristics:

(1) marked impairment in the use of multiple nonverbal behaviours such as eye-to-eye gaze, facial expression, body postures and gestures to regulate social interaction

(2) failure to develop peer relationships appropriate to developmental level

(3) lack of spontaneous seeking to share enjoyment, interests or achievements with other people (i.e., by a lack of showing, bringing, or pointing out objects of interest to other people)

(4) lack of social or emotional reciprocity

B. Restricted, repetitive and stereotyped patterns of behaviour, interests and actions manifested by at least one of the following characteristics:

(1) encompassing preoccupation with one or more stereotyped and restrictive patterns of interest that is abnormal either in intensity or focus

(2) apparently inflexible adherence to specific, non-functional routines or rituals

(3) stereotyped and repetitive motor mannerisms (i.e., hand or finger flapping or twisting, or complex whole-body movements)

(4) persistent preoccupation with parts of objects

C. The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.

D. There is no clinically significant general delay in language (i.e., at 2 years of age using simple words, at 3 years communicative phrases are used).

E. There is no clinically significant delay in cognitive development or the development of self-help skills appropriate for the age, adaptive behaviour (other than social interaction) and curiosity about the environment in childhood.

F. Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia.

Diagnosis of F84.9 Pervasive Developmental Disorder not otherwise specified (including atypical autism) [299.80

This category should be used when there is a severe and pervasive impairment in the development of reciprocal social interaction or verbal and nonverbal communication skills, or when stereotyped behavior, interests, and activities are present, but the criteria are not met for a specific Pervasive Developmental Disorder, Schizophrenia. For example, this category includes "atypical autism", cases that do not meet the criteria for Autistic Disorder because of late age of onset, atypical symptomatology, or subthreshold symptomatology, or all of these.

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Summary of evidence

II, III	The diagnostic criteria of DSM-IV-TR and ICD-10 increases the reliability of the diagnostic process, especially in inexperienced professionals[149-151]
	The diagnostic criteria of DSM-IV-TR and ICD-10, alone, have little discriminatory validity for the Asperger disorder and autism[149-151]

Recommendations

C	It is recommended that professionals involved in the detection of children with ASD, especially those with little experience, use the diagnostic DSM-IV-TR and / or ICD-10 manuals

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6. Early Detection

The questions that are going to be answered in this chapter are:

- Why is early detection important?
- How should the monitoring of a healthy child in Primary Care be to detect problems early in his or her development?
- Are there any useful warning signals / criteria for specific suspicion of ASDs for early detection in Primary Care?
- May the parents' warning be effective in Primary Care for risk assessment of ASD?
- What rating scales of suspected ASD are most effective?
- Do we need a more comprehensive monitoring in risk groups?
- What is the minimum age of suspicion?

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6.1. Importance of Early Detection

From an evolutionary perspective, early intervention seems to improve overall prognosis of children with ASD in terms of adaptability in the future, together with added benefits for the family, although the investigations carried out in this regard are limited in number and methodological quality. Some studies have shown positive results especially in relation to problems in communication and social interaction, although more studies are still necessary[33,152].. Therefore, although controlled trials are needed which compare the same intervention performed early and performed at older ages, it is undeniable that early intervention, which provides adequate supports (individual, educational, family and social) largely affects the quality of life of the children as well as the coping capacity of families, facilitating their future social inclusion as an independent person, favouring positive results in areas of cognition, language and daily living skills,[153.154].

Some studies have identified those variables that are predictive of positive outcomes in children diagnosed with autism not associated with intellectual disabilities and Asperger disorder. These variables are mainly the language and nonverbal skills. The predictive power of these variables supports early intervention programs, especially in regard to language[46,155].

Cohort Study (2 +)

Another study explored age and IQ before and after an intensive treatment program, determining that there is a significant relationship between the child's age when admitted and his or her educational situation at the end, this being the best among those who were younger when they entered. The IQ also proved to be a predictor of the final educational status[156].

Cohort Study (2 +)

There are justifications for early intervention based on knowledge of neuro-plasticity. Although it is not yet known how to capitalize fully brain plasticity and its potential, it is agreed that the clearest early stimulation techniques are those following the way of nature, facilitating the normal processes of development based on interventions with the child, family and the environment[152].

Expert opinion (4)

Another important aspect in favour of early detection, given the significant genetic component of ASD, is the genetic counselling that can be offered to families when ASD is detected in a child at an early stage. This advice is just as important as the intervention of the affected child. However, genetic counselling will vary depending on certain factors (whether it is primary or secondary autism and whether there are gene causes)[33,157-159].

Descriptive Studies (3)

Summary of evidence

2 +	Early detection of children with ASD allows early intervention and leads to better performance results particularly in children with autism not associated with intellectual disability and among those who were younger when admitted[46,155]
3, 4	Early detection allows genetic counselling of families because of the significant genetic component of ASDs[33,157-160]

Recommendations

C	Early detection of children with ASD is recommended as part of the healthy child care process

6.2. Follow-up of a healthy child for early detection of ASD

Autism is seen as a problem to be taken into account in the monitoring of a healthy child[161-163], however, training on this matter is scarce and the use of developmental scales is not standardised. In some cases, these scales have low sensitivity and specificity to detect certain development problems, as is the case of Devopmental Denver Screening Test (DDST-II), the monitoring test that is most used at a global scale[164,165].

Denver Scale-II (DDST-II) from 1992[166] is a revision and update of the Denver Developmental Screening Test, DDST (1967)[167], which explores four areas of development (fine motor, gross motor, personal-social and language) from 3 months to 6 years of age. According to other authors, it has a 43% of sensitivity, a specificity of 83% and a positive predictive value of 37%[168]. A study reviewed the more standardised scales used to assess child development, discarding the Denver-II test because of its low sensitivity and specificity[165].

Diagnostic studies (III)

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In our context, the most widely used scales for the monitoring of healthy child development are the Denver Developmental Screening Test (DDST-II) and the Haizea-Llevant scale[169.170]. The latter has been developed in our country from a review of the items used in different scales used internationally such as a Denver-II, Bailey, and others.

Descriptive study (3)

The Haizea-Llevant scale checks the level of cognitive, social and motor development in children from 0 to 5 years of age. This instrument was developed with a representative sample of 2519 children in the Autonomous Communities in the Basque Country and Catalonia. It offers the normal acquisition range of some basic skills in childhood. This test includes 97 items distributed in the following areas: socialisation (26 items), language and logical-mathematical (31 items), handling (19) and postural (21 elements). Each of the elements indicated the age at which 50%, 75% and 95% of the children performed it. In addition, as an added value, the instrument includes some warning signs, whose presence, at any age or from specific ages, indicates the possibility of alterations.

The CPG of New Zealand (NZ) 2008[46] based on the opinion of the experts who conducted the Manual on healthy children of the Ministry of Health from New Zealand in 1996[171] describes what should be the portfolio of services for the monitoring of a healthy child to identify any problems in its development, taking into account areas of valuation, education / promotion activities and moments for parents to develop skills on the monitoring and care of children. The authors establish monitoring procedures and follow-up of child development, by exchanging information with parents and caregivers taking advantage of every encounter or visit.

CPG Expert opinion (4)

The NZ guide sets out how health and educational professionals should regularly discuss the development of children with parents (at least between 8-12 months, 2-3 years and 4-5 years) as part of the monitoring of the healthy child to detect and respond to any problems promptly[46,171].

The CPG of SIGN-2007[148] presents a study, which evaluates medical histories of children with ASD born between 1991 - 1992 to see how the detection process was performed. The estimated prevalence rates were high, 55% more compared to the results for the period 1991-96. Experts believe that the detection of cases has been higher in the last decade due to the level of vigilance and not a real increase in cases. It is believed that the incorporation of a high level of vigilance by professionals in the fields of social, play, language and behaviour development, improves the early identification of ASD and other disorders.

Descriptive study (3)

CPG Expert opinion(4)

Summary of evidence

4	Health professionals establish certain procedures for monitoring and follow-up of child development, exchanging information with parents and caregivers taking advantage of every encounter or visit[171].

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4	The New Zealand guide states that health and educational professionals should regularly discuss the development of children with the parents (at least between 8-12 months, 2-3 years and 4-5 years) as part of the monitoring of the healthy child to detect and respond to any problems promptly[46,171]
3, 4	The incorporation of a high level of vigilance by professionals in the fields of social, play, language and behaviour development, improves the early identification of ASD and other disorders[19,148]
III, 4	There are development monitoring scales used in our context as the Haizea-Llevant scale (developed in our context) and the Denver-II (not validated in our context). The latter has been ruled out by international experts for its lack of sensitivity and specificity[166,169]

Recommendations

D	Health and educational professionals should regularly discuss with parents the psychomotor, intellectual and behavioural development of their children (at least between 8-12 months, 2-3 years and 4-5 years) as part of the monitoring of the healthy child
D	Health professionals should incorporate a high level of vigilance in the fields of social, play, language and behaviour development for the early identification of ASD and related disorders
√	In the monitoring of healthy child development in Primary Care, to help detect any developmental disorder, including ASD, we recommend the use of scales (such as the Haizea-Llevant scale developed in our context) (Appendix 3)

6.3. Warning Signs and Scales

6.3.1. Warning Signs

The knowledge of what is normal development for both parents and professionals is the first step when identifying the problems. Similarly important is the fact of knowing the family history as well as the perinatal background, the development in the acquisition of the skills expected by the age of development: "child development milestones"[172]

There are some immediate warning signals for any child which indicate the need for a broader and more specific assessment[172]: Expert Opinion (4)

- Does not babble, does no gestures (pointing, waving good-bye) at 12 months.
- Does not say single words at 18 months.
- Does not say two-word spontaneous phrases (not echolalic) at 24 months.
- Any loss of language skills or social level at any age.

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There are other warning signs of child development, important when there is suspicion of ASD, based on the consensus of the development group of this guide following the opinion of other international experts46,[172-175]. These signals are presented according to their most frequent appearance by age groups, considering that the signs of previous stages can appear at any age. These signals are explained in the following table (Table 6 and Annex 4):

Expert Opinion (4)

Table 6: Summary of ASD warning signs by age periods

Note: <u>The signs established in previous stages can appear at any age.</u> These factors alone do not indicate ASD. They should warn professionals on the risk of ASD, so that the referral depends on the situation as a whole. They cannot be interpreted from the evolutionary point of view of ASD either, but as potential signs that can be found in children evaluated at the ages referenced.
Before 12 months
– Low frequency of use of gaze directed at people – Shows no advance when going to be held – Lack of interest in simple interactive games like “peek-a-boo” or “ give and take “ – Lack of social smile – Lack of anxiety to strangers when about nine months
After 12 months
– Less eye-to-eye gaze – Does not respond to name – Does not point to “ask for anything” (proto-imperative) – Does not show objects – Unusual response to auditory stimuli – Lack of interest in simple interactive games like “peek-a-boo” or “ give and take “ – Does not look where others point out – Lack of spontaneous imitation – No social / communicative babbling as if it talked with adults
Between 18-24 months

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– Does not point the finger to “share an interest” (protodeclarative) – Has difficulty to follow the gaze of an adult – Does not to look where others point out – Delayed development of understanding and / or expressive language – Lack of functional play with toys or presence of repetitive forms of play with objects (i.e., put in line, open and close, switch on and off, etc) – Lack of symbolic play – Lack of interest in other children or siblings – Does not usually show objects – Does not respond when called – Does not imitate or repeat gestures or actions that others do (i.e., grimaces, claps) – Few expressions to share positive affect – Used words before but not anymore (language regression)
From 36 months

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Communication

– Absence or language delay or deficit in language development is not balanced by other modes of communication

– Stereotyped or repetitive use of language such as echolalia or refer to himself in 2nd or 3rd person

– Abnormal intonation

– Poor response to its name

– Deficits in nonverbal communication (i.e., no pointing out and difficulty identifying and sharing a "focus" with the look)

– Failure of social smile to share enjoyment and respond to the smiles of others

– Achieves things by himself, without asking

– Used words before but not anymore

– No varied representation games or social imitation appropriate to developmental level

Social disorders

– Limited imitation (i.e., clapping) or absence of actions with toys or other objects

– Does not "show" objects to others

– Lack of interest or strange approaches to children of its age

– Little recognition or response to happiness or sadness of others

– Does not join others in shared imagination games

– Failure to initiate simple games with others or participate in simple social games

– Preference for solitary activities

– Strange relations with adults from an excessive intensity over to a striking indifference

– Lack of social gaze

Disorders of interest, activities and behaviours

– Insistence on routines and / or resistance to changes in unstructured situations

– Repetitive games with toys (i.e., align objects, turning lights on and off, etc)

– Unusual attachment to a toy or object which it always carries that interferes with its daily life

– Hypersensitivity to sounds, touch and certain textures

– Unusual response to pain

– Unusual response to sensory stimuli (auditory, olfactory, visual, tactile and taste)

– Strange postural patterns such as tiptoe

– Motor stereotypy or mannerisms

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Screening from 5 years *
Communication Disorders – Poor language development, including muteness, odd or inappropriate intonation, echolalia, unusual vocabulary for its age or social group – In the case where there are no deficiencies in language development, there is limited use of language to communicate spontaneously and a tendency to speak only about specific topics of its own interest (fluid language but hardly appropriate to the context) **Social disorders** – Difficulty joining the games of other children or inappropriate attempts to play together – Limited ability to appreciate cultural norms (in dress, style of speech, interests, etc.) – Social stimuli will produce confusion or displeasure – Inappropriate relationships with adults (too intense or nonexistent) – Shows extreme reactions to the invasion of its personal or mental space (intense resistance when to much pressure is put with different instructions to its focus of interest) **Limitation of interests, activities and behaviours** – Lack of flexibility and cooperative imaginative play, but usually creates alone certain imaginary scenarios (copied from videos or cartoons) – Difficulties of organisation in unstructured spaces – Inability to deal with changes or unstructured situations, including those, which the children enjoy such as school excursions, when the teacher is absent, etc. – Collects data on certain topics of interest in a restrictive and stereotyped way **Other features** – Unusual abilities and weaknesses profile (i.e., poorly developed social and motor skills, gross motor clumsiness) – General knowledge, reading or vocabulary may be beyond the chronological or mental age) – Any significant history of loss of skills – Certain areas of knowledge may be particularly strong, showing amazing skills in areas such as mathematics, mechanics, music, painting, sculpture
* The warning signs from 5 years of age are more focused on those cases that may have been overlooked in previous assessments, as they were ASD conditions with less impact as the Asperger case.

Summary of evidence

4	The work of early identification of ASD by health professionals is made easier by taking into account the "milestones and warning signs in the development" of the healthy child[46,172-175]

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Recommendations

D, √	Healthcare professionals should monitor the development of healthy children taking into account the "warning signs" proposed: – Immediate warning signals – Compendium of ASD warning signs according to age periods

6.3.2. Parental concerns (warnings from parents)

According to a UK study of 2001[176], in most cases (60%) parents are the first to suspect the problem compared with 10% of health professionals and 7% of education professionals. The New Zealand CPG-2008[46] also refers to the previous descriptive study and to another from 1998 also from the United Kingdom[177], which contains information on families of children affected by ASD, and it identifies the needs of children, as well as their families, and which explores the vision of the parents upon the diagnostic process of their children (age at diagnosis, delay problems, practitioners involved, etc). — Descriptive Studies (3)

Other descriptive studies[148,178-180] which explore the initial clinical and diagnostic process of children with ASD yield similar results while the parents are usually the first concerned, especially in aspects of language development, followed by the presence of abnormal socio-emotional responses and medical problems or developmental delay. — Descriptive Studies (3)

In a review performed on 78 published studies, which compared the information given by parents to that of paediatricians, it was found that in most of the studies the rates observed were between 75% and 95% of the times when there was agreement between the observations of both[181]. — Expert opinion (4)

Two descriptive studies explored the predictive value of the concerns of 95 parents about the behaviour of their children completing a sensitivity of 70% and a specificity of 73% for such concerns[182]. — Diagnostic study (III)

Another study explored the parents of 234 children from birth to 77 months to assess whether the parents could adequately overestimate age compared with chronological development of their children, and found a sensitivity of 75% if it was less than the chronological age and a sensitivity of 90% if the developmental age was less than the chronological one[183]. — Diagnostic study (III)

In our context, a research with nearly 650 families across the Spanish territory (2003-04)[184] has been carried out, collecting data on the diagnostic process followed by those families with a child with autism. — Descriptive study (3)

The results of the study indicate that in 80% of cases the family is the first to suspect that there is a problem in the development of their child, with an average age of suspicion one year and ten months for children with PDD. Only 5% of the families say that the paediatrician was the first to suspect a developmental disorder, though, when he or she does, it is at an average younger age than when these suspicions are observed by a member of the family.

More than 5 years have passed since the publication of this Clinical Practice Guideline and its validity has not been reviewed

There are some studies that assess the ability of a tool developed on parental concerns: the Parent's Evaluation of Developmental Status (PEDS) scale. This tool is self-administered to the parents, has a sensitivity rate between 74% and 79% and a specificity of 70% -80% to detect developmental problems of children from 0 to 8 years old. This tool has been translated but it has not been validated in our country[185,186].

Diagnostic study (II)

Summary of evidence

3	Although parents usually suspect something is wrong with the development of their children when they are around 18 months of age, the diagnosis is made later[148,176 – 180,184]
III, 3	Parents play an important role in monitoring the development of their child as frequently they are the ones who detect in most cases that something is wrong with their children[148,176-178,180-184]
II	The PEDS scale on the concerns of the parents has a sensitivity of 74% -79% and a specificity of 70% -80%, although not used as a screening tool, it can help to identify children suspected of having a developmental problem in Primary Care[185.186]

Recommendations

C	Concerns or warnings from parents regarding their children's development should be valued as much as the very presence of abnormal features
C	Take advantage of any visit (routine monitoring or due to illness) to consult the parents on concerns regarding their child's development
√	Professionals can use the items of the PEDS scale to direct questions about parental concerns (Appendix 5)

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6.3.3. Early screening scales

There are different instruments used to detect autism spectrum disorders; there is a Spanish version of them, although it has not been validated, the Checklist for Autism in Toddlers (CHAT)[187] by Baron Cohen et al., designed to discriminate adequately between children with normal neurodevelopment and children with autism spectrum disorder before two years of age. Since this instrument has a high specificity (99%) but low sensitivity (38%) it is not recommended as a screening method. In fact, the authors are validating an enhanced version, the Q-CHAT[188].

Diagnostic study (II)

There is an additional scale, the Modified Checklist for Autism in Toddlers (M-CHAT)[189,190], designed to increase the sensitivity of the CHAT for detecting ASD in children between 16-30 months of age. It consists of an extension of the CHAT instrument to 23 questions (previously there were 9 questions) self-administered for parents. The authors considered altered result criteria when failing 3 of the 23 questions or failing 2 of the 6 questions considered key: interest in other children and imitation, joint attention (use of protodeclaratives and eye tracking), bringing objects to show parents and answer when called. This change increased the sensitivity and specificity to 97% and 95% using the first criterion, and to 95% and 99% using the second criterion, respectively.

Diagnostic study (II)

This tool has been translated into Spanish and it is validated and about to be published in a study led by the Spanish group GETEA. It is a longitudinal, population-based study, with a total of 2480 children between 18 to 24 months of age, from the areas of Salamanca and Zamora, who came to the consultation of a healthy child and / or vaccination. The results estimated a sensitivity of 100% and a specificity of 98.3% to discriminate between children with ASD and children without ASD, and a sensitivity of 75.3% and 99.8% specificity to discriminate between children with ASD and children with other disorders (data provided by the research team).

Under the concept of mass screening tool, experts from the United Kingdom consider[148.191] that high sensitivity levels are necessary, as the ones existing currently do not meet the instruments of the CHAT and M-CHAT. However, the M-CHAT can be used as an aid instrument to identify ASD suspicious risk factors.

Expert opinion (4)

A systematic review[192] from 2005 of English screening instruments for autism discards CHAT as a screening instrument and recommends further investigations on the competences of the M-CHAT.

Descriptive study (3)

The SIGN guide148, the AAP (American Academy of Paediatrics) 193 and the development group of this guide have regarded M-CHAT as a useful tool for confirming clinical suspicion of ASD.

CPG Expert opinion (4)

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There is another questionnaire developed for ASD screening, the Social Communication Questionnaire (SCQ). It has been developed to assess children older than 4 years old with a mental age of over 2 years. It is available in two forms of assessment of the current situation or lifelong. There are 40 questions to be answered with YES or NO and it is self-administered by the parents. This questionnaire is translated into Spanish, but it has not been validated in our context. It was performed on a population of 160 individuals with PDD and 40 without PDD between 4-5 years of age, giving a sensitivity of 85% and a specificity of 75% from a cut-off of 15. Further studies show very similar results: 86% sensibility and 78% specificity for a population of 119 children between 9-13 years with special education needs and without ASD. Another study showed an 88% sensitivity and 72% specificity with children between 9-10 years within a cohort. In addition, in another study on 81 children aged between 2-6 years the data varied: sensitivity 93% and specificity 58% for children aged between 2-6 years and sensitivity 100% and specificity 62% for children between 3-5 years with a cut-off point of 11[194-198].

Diagnostic study (II,III)

Within the specific detection of Asperger disorder, we find tools such as the Childhood Asperger Syndrome Test (CAST) scale, which screens people with Asperger disorder. There are 39 questions to be answered with YES or NO and it is self-administered by parents. This tool was developed in a pilot study with a total of 50 children between 3-9 years with Asperger disorder or autism and children with an absolutely normal development. The validation was conducted on a sample of 139 children aged between 4 and 11. The estimated sensitivity was 87.5% and specificity 98% for a cut-off point of 15 for cases of Asperger's Disorder[199]. A study obtained higher data but on a minor sample[200]. Another study assessed the test-retest reliability[201] with a sample of 73 parents who completed the CAST test twice in two months after the first time, giving a Kappa of 0.41 and a Spearman Correlation Coefficient of 0.67. The scale has not been validated in our context.

Diagnostic study (II)

Descriptive study (3)

There is also a scale that has been developed and validated in our context, the *Autonomous Scale for detection of Asperger's disorder and high functioning autism*[202]. It is a scale of 39 questions to answer from five categories (never, sometimes, often, always, not observed) self-administered by parents and teachers. The validation process has been conducted on a population of 109 people aged between 5 and 31 in three groups: one composed of children and youth diagnosed with Asperger's disorder, another of autism not associated to intellectual disability, and another disorder with attention hyperactivity deficit. The results estimated sensitivity between 97.8% and 100% and specificity between 98.9% and 72% for the 37 and 36 cut-off points respectively, with a 30.2% of false positives in cut-off point 36. The authors suggest referring the child as from cut-off point 36. The interrater correlation (0.83) and test-retest (0.94 for parents and 0.97 for teachers) gave positive results.

Diagnostic study (II)

More than 5 years have passed since the publication of this Clinical Practice Guideline, and its validity has not been reviewed

Summary of evidence

4	No instrument has been identified for the detection of ASD which meets the criteria for population screening test[148.191]
2	The Checklist for Autism in Toddlers (CHAT) scale has a specificity of 99% but a low sensitivity of 38%. The M-CHAT amended version has a better sensitivity of 95% and a specificity of 99%, although more studies on their psychometric properties are needed. The validation data in Spain offer a 100% sensitivity and specificity of 98.3%[148,187,187,189,67,190,192]
4	The SIGN guide 148, the AAP (American Academy of Paediatrics)[193] and the development group of this guide have regarded M-CHAT as a useful tool to confirm clinical suspicion of ASD in children between 16 and 30 months.
2	The Autonomous scale for the early detection of Asperger's disorder has improved from 97.8% to 100% sensitivity and specificity between 72% to 98.9% from a cut-off point of 37, although more studies on its properties in a wider population are needed[202]

Recommendations

C	Population screening for ASDs is not recommended with the instruments developed at this moment, although the capability of the M-CHAT and the Autonomous Scale is being investigated
B, C	We recommend the M-CHAT instrument as a useful tool to confirm clinical suspicion of ASD in children between 16 and 30 months (Appendix 6)
B, C	We recommend using the Autonomous Scale, in children older than 5 years as a useful tool to confirm clinical suspicion of Asperger's disorder (Appendix 7)

6.3.4. Monitoring risk groups

The monitoring of children at high risk is referred to the assessment of children with ASD who due to their personal or family history suffer a greater risk of ASD, although no specific symptoms have been observed yet.

Cohort study (2+)

According to studies on the aetiology of ASD, it has been stated that children who have presented a process which could have affected the perinatal nervous system (such as maternal consumption of alcohol, teratogenic drugs or other substances during pregnancy, or even pre-or postnatal infections such as congenital rubella, haemophilus influenza and cytomegalovirus, that may involve meningitis or encephalitis), could be more likely to develop ASD. In addition, some metabolic diseases such as PKU not treated after birth. The presence of epileptic seizures (in the context of epileptic encephalopathies) in the first year of life also appears to be associated with later diagnosis of autism, although it cannot be considered a risk factor but associated comorbidity[128]. Other observational studies have found that older age in the father or mother is associated with risk of ASD; however, the presence of hyperbilirubinemia at birth is not a risk factor[203,204].

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In another observational study, the main results suggested that perinatal factors such as breech presentation, low Apgar score (≤ 7 at 5 min.), low birth weight, birth before 35 weeks and small size for gestational age, significantly increased the risk of ASD. Likewise, children of parents with psychiatric history of schizophrenic psychosis or affective disorder also had an increased risk of ASD. These factors appeared to act independently[205].

Cohort study (2+)

A cohort study on a population of children between 3 and 10 years in Atlanta identified 617 children with autism. After adjusting to the birth history data it showed that a birth weight <2500g and preterm <33 weeks was associated with an increase of two times the risk of autism, although the magnitude of risk varied according to gender (higher in girls) and the subgroup of autism (higher for autism accompanied by other disabilities such as intellectual disability)[206].

RS from Cohort study (2++)

A review of seven studies (four cohort and three case-control) on ASD risk factors concluded that the studies suggest that parental age and certain obstetric conditions are associated with an increased risk of ASD, but are not tested as independent factors for ASD. These variables should be examined in future studies with a cohort population, as some results are inconsistent. Genetic susceptibility to autism, as a clear risk factor for ASD, which is itself associated with sub-optimal obstetric conditions, should be taken into account as a potential prospect for future analysis[207].

RS from Cohort study (2++)

Another systematic review with meta-analysis evaluated factors related to prenatal risk of ASD. Factors positively associated with risk of autism were older parents and complications during pregnancy (gestational diabetes and bleeding). However, it concluded that there was insufficient evidence to implicate any of the prenatal factors in the aetiology of autism[208]. A prospective study that evaluated parental age (not included in the review above) reached the same conclusion after adjusting the analysis to a maternal age ≥ 35 years versus 25 to 29 years; paternal age ≥ 40 years versus 25-29 years, concluding that parental age was positively associated with risk of autism[209].

Cohort study (2+)

Descriptive Studies (3)

Due to the genetic component of ASD, those siblings of children diagnosed with ASD have a higher probability of developing ASD than the rest of the general population, which may be even higher, if the genetic component is determined as in the case of some associated syndromes. These children with relatives suffering from ASD (parents or siblings) are candidates for a more comprehensive monitoring by Primary Care professionals.[60,66,67,74,157-160,210].

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Summary of evidence

2+ 2++	There are certain factors that appear to increase the risk of development of ASD: perinatal factors (consumption of teratogenic drugs or substances, perinatal infections, gestational diabetes, gestational bleeding, foetal distress, prematurity, low birth weight), metabolic diseases, older parents and parents with a history of schizophrenic or affective psychosis[128.203 - 207.209].
3	The genetic component of ASD makes those siblings of children diagnosed with ASD have a higher probability to develop ASD than the rest of the general population[60,66,67,74,157-160,208,210]

Recommendations

C, D	It is recommended to take into account those factors associated with the development of ASD in regular visits: perinatal factors (consumption of drugs or teratogenic substances, perinatal infections, gestational diabetes, gestational bleeding, foetal distress, prematurity, low birth weight), metabolic diseases, older parents and parents with a history of schizophrenic or affective psychosis and a family history of ASD (siblings with ASD)
C	Professionals should carry out a close monitoring of the development of those siblings of children with ASD as these are a risk group

6.3.5. Minimum age of suspicion

Although specialists tend to detect this disease from two years of age, the manifestations may be seen before. The studies that gather the concerns of parents report that they usually realize at around 18 months of age that there is a strange behaviour in their children, many even before[174].

It is complex to determine exactly when changes are evident and what specific changes other than the behaviours of a child without manifest disorders, as studies that can determine the signals are retrospective and based on observation and home videos as well as on the exploration of what parents remember, so the risk of obtaining biased results is higher. There is another group of longitudinal studies, which monitors younger siblings of children diagnosed with ASD, whose chances of having ASD are also higher and which can perform a more detailed observation and recording of their behaviour. Other studies reaffirm that there is significant stability in the diagnosis of ASD if made before the age of three[211].

The studies found no evidence of a minimum age for specific diagnosis of ASD, but early manifestations may occur at very early ages, before the two years of age, although it must be taken into account that the lack of such behaviours does not rule out the possibility of presenting subsequent alterations[148].

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Another double-blind case-control study has evaluated the behaviour in home videos (from the first 6 months of age) of children diagnosed with ASD before the age of 3 years versus the observation of videos of a group of children without ASD. The study indicates a high interobserver concordance. The results established that there were significant differences in frequency between the two groups for manifestations such as: looking at people, targeting people, seeking contact, smiling at people and vocalizing at peopl[212].

Cases and Control (2 +)

In the cohort study previously described in the follow-up section of children at risk, a sample of 150 siblings of children with ASD, various disorders in the first year of life are identified, in the group of children later diagnosed with autism, both in the fields of social and non-social development[26]. Other prospective studies assessed the stability of the diagnosis of ASD made at 2 and 9 years old. The concordance percentage between the diagnosis of ASD at 2 years and 9 was 67%[213,214].

Cohort Study (2 +) Descriptive Study (3)

Another prospective longitudinal study assessed the behaviour of children included in the follow-up of 3021 children less than two years that had not been previously identified with any developmental delay. There were archival videos from the first year of age of a group of 18 children later diagnosed with ASD to a group of 18 children with other developmental disorder not ASD and against another group of children without any developmental disorder. Additionally, the data included periodic examinations and parental responses to the question of whether they were concerned about their children's development.

Cases and Control (2 +)

There were some differences among the group of children with ASD compared with the group with other developmental disorders and children with ASD with the group of children with no disorder: no social gaze, eyes that express a lack of affection and closeness, lack of response to their name, lack of shared enjoyment or interests, no pointing out and present body and object stereotypias. There were no differences between the group of children with ASD and the group with other disorders, but there were differences with respect to the group without disorders: lack of anticipatory movements or positions, failure to respond to contextual cues, lack of vocalisations with consonants, absence of functional game with toys and difficult to calm down when they get upset[243].

A study explored the children's behaviour through in-depth interviews of what parents remembered about the behaviour of their children at a young age[214]. A review of studies evaluating the warning signs from an early age concludes that: changes are evident before 12 months and that 80% of parents noted disorders before 2 years of age with an average of 18 months[211]. Another study evaluating home videos on a group of children with ASD compared to another without ASD at one year old, found different behaviours in both groups[216].

Descriptive Study (3)

Cases and Control (2 +)

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Summary of evidence

2+ 3	There are significant differences in the behaviour of children with ASD observable from 6 months old in comparison to the behaviour of children without disorders. As the months pass, the differences in development with respect to children without ASD, in both social and non-social fields, become more noticeable. There is a significant percentage of diagnostic stability after 2 years of age[26, 211 - 216]
2+ 3	There is no conclusive evidence of which is the minimum age to detect ASD disorders[26, 211 – 213.215]

Recommendations

C	Practitioners should monitor the presence of any warning signals from at least 6 months of age
√	Given that there is no evidence of a minimum age for alterations, referral to Specialised Care may be considered at any age when there is a suspicion of ASD

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7. ASD suspicion management strategies in Primary Care

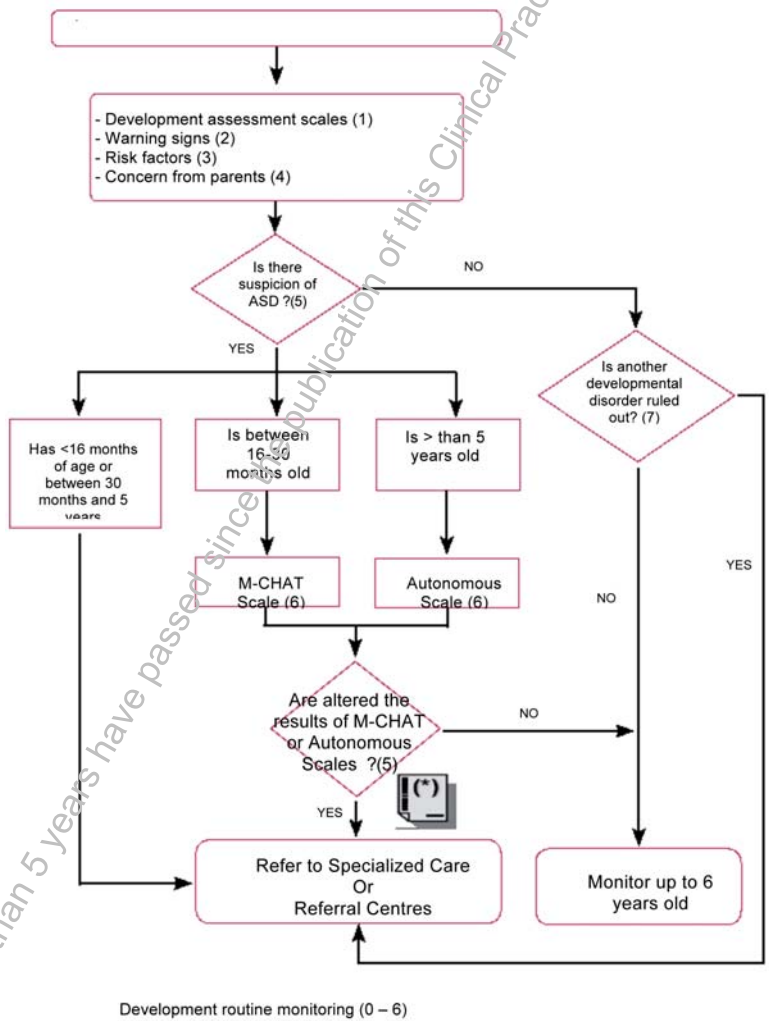
The questions that are going to be answered in this chapter are:

- What are the steps to follow if there is any suspicion of ASD? - Algorithm
- What are the referral criteria of ASD? - Algorithm

Steps to be taken if there is a suspicion of ASD

As initial guidance, in a patient with symptoms of ASD, the following algorithm is presented to guide the referral work, based on the recommendations made earlier in the guide:

ALGORITHM FOR THE TREATMENT OF SUSPECTED ASD IN PRIMARY CARE



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NOTES ON THE MANAGEMENT ALGORITHM OF ASD SUSPICION IN PRIMARY CARE:

(1) Developmental assessment scales: Haizea- Llevant Scale (developed in Spanish context). (Annex 3)

(2) Immediate warning signals and compendium of ASD warning signs by period of age (Annex 4)

(3) ASD risk factors to be considered when assessing the child's background: perinatal factors (consumption of drugs or teratogenic substances, perinatal infections, gestational diabetes, gestational bleeding, foetal distress, prematurity, low birth weight) metabolic diseases, older parents, parents with a history of psychosis or affective schizophrenia and family history (siblings with ASD)

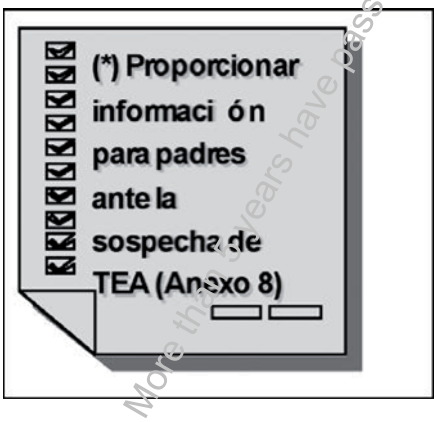
(4) The concern of parents. Paediatric Emotional Distress Scale (PEDS) to guide the professional about what to ask parents (Annex 5)

(5) Discard metabolic and hearing disorders

(6) M-CHAT and Autonomous Scale. Use as a supplement if there is a suspicion in children:

- Between 16-30 months: M-CHAT (Self-administered to parents) - Annex 6
- Over 5 years: Autonomous Scale (Self-administered to parents and teachers) - Annex 7

(7) Other developmental disorders. It refers to the presence of other disorders that do not meet the criteria of ASD, such as motor skills disorders (for example ataxia, poor coordination, hypotonia that are indicative of Infantile Cerebral Palsy, etc.) intellectual disability (any grade), learning disorders, or communication disorders (expressive, receptive, mixed or phonological) and others.



Provide information to parents if there is a suspicion of ASD (Appendix 8)

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8. Keys/Aspects to consider when providing information to parents

The questions that are going to be answered in this chapter are:

- Does the information given to parents of children with suspected ASD amend their satisfaction and their ability to cope with the problem?
- What are the aspects to consider in relation to parents when providing information if there is a suspicion of ASD, and after a diagnosis of ASD?
- What is the basic information that Primary Care professionals are required to provide to parents of children with suspected or newly diagnosed ASD?

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8.1. Aspects to consider when providing information to parents for suspected ASD

Inform a parent that their child may have a problem in its development is one of the most difficult news to transmit, so we must be careful when providing that information.

We can find two situations at the visit. The first when the parents already suspected that her son had any problems, so the confirmation by the professional is experienced as a relief as it reaffirms their suspicions. In such a case, the referral process is welcomed as a positive thing because parents are grateful that something is done accordingly and inquire about the problem.

The second situation that we can find is that parents first hear that their children may have a developmental disorder, so the way to act must be more cautious. He or she is to be referred too, but making parents aware that there is a problem, which ought to be investigated. This situation may be more common in first-time parents in which there is no previous comparative reference. The professionals provide information on what are normal developmental stages in a child from birth; therefore, it is as important as any other information given to them on the care of their child.

Sometimes, when the first warning signs are identified by parents, these may hesitate to deny or accept this reality. "Turning a blind eye" to this reality is a self-defence mechanism against the anxiety that it produces.

The moments experienced by the parents in the face of evidence that there is a disorder and getting a diagnosis, result in high levels of stress and anxiety due to uncertainty. Some studies describe how in many cases, when consulted with professionals such concerns, they denied or played down their importance, thereby contributing to increased stress and feelings of helplessness, and devaluating the trust in health professionals[217.218].

During the prediagnostic period, feelings of stress and confusion require support by professionals. The time factor is very important; the confirmation of a diagnosis will decrease the uncertainty of the process[217].

Qualitative Research

A phenomenological qualitative study described the process of raising a child with autism, discussing the diagnosis process and the influence of autism on different areas of the family. The main experiences on the process of diagnosis are as follows[219]:

- Some participants reflected the fact that they had bad experiences in the early demands of professional help. Participants felt that the professionals refused their concerns and did not pay the necessary attention to the delayed development of their children, which contributed to increased stress and feelings of powerlessness and helplessness.
- Reflected difficulty to detect signals from first-time parents.
- Parents often denied or downplayed the early signs they observed which implied that something is wrong with their child's development, before asking professionals.
- The time between the early warning signs of autism and the definition of the diagnosis were the most difficult in their lives with their children with ASD. They often felt blocked, anxious and scared at the prospect that something could be terribly wrong with their children, as well as exhausted by their inability to understand the behavioural problems of their children. They experience the diagnostic process as a period of stress and great uncertainty.
- When finally a diagnosis is found, at the beginning there can be a relief because it reduces the uncertainty lived during the process.
- The participants did not see anything strange in their babies; they were healthy babies growing well, so the diagnosis is received with great shock.
- Some participants experienced feelings of guilt because they felt responsible for what was happening to their children or because they failed to react to the strange behaviour of their children.

Descriptive Study (3)

A descriptive study explored the satisfaction determinants of parents during the diagnostic process through a questionnaire self-administered to 126 parents (73% mothers and 27% fathers) of children with autism. This study concluded that there was a significant association between the levels of parental satisfaction and items such as: suspicions of parents considered by professionals, the possibility to ask questions when they are given the diagnosis, the quality of the information received and professional manners. 35% were very satisfied on how they were given the diagnosis, 33% satisfied, 7% were dissatisfied and very dissatisfied 6%[218].

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Qualitative research

A qualitative study explored the perceptions of parents on the process of communication with professionals during the diagnosis. 15 focus groups of parents with children with ASD whose diagnosis was detected a maximum of 7 years ago to a minimum of six months ago were conducted. Among the key findings[220]:

- 30% of participants did not receive additional information or advice or support on the nature of ASD when they received the diagnosis.
- Many parents suggested that communication with professionals could be improved if they were suggested how to contact support groups or parent groups, and if there was a possibility to get in contact with Specialised Care professionals in an easier and more common way to address the different questions which arise.
- Parents called for better training and guidance regarding ASD for professionals.

Descriptive study (3)

A descriptive study explored the importance of knowledge of ASD among health professionals through a questionnaire conducted on 146 primary caregivers of children with ASD, including the demographic and diagnostic process data, sources of information, support and perceived needs. Among the most relevant data the following should be taken into consideration[221]:

- Between 71-73% of primary carers relied on media sources (Internet, books, videos, etc) for information, 42% on conferences / workshops and 42% on other parents. Only 15-20% resorted to health or educational professionals. 64% reported they belonged to a parents' association.
- Most professionals, 82% gave some additional information at the time of early diagnosis, compared to 18% who did not provide either additional information or referred to specialist services.

Summary of evidence

Qualitative Research	– Some parents report that some professionals refused their concerns and ignored them in relation to the developmental delay of their children, which increased the stress and sense of helplessness – New parents can prove difficult to detect warning signs because they have no prior references – The diagnostic process is experienced as a period of stress and great uncertainty – The diagnosis is received at the same time as a relief, since it ends the uncertainty, and as a great shock by the reality that is presented. The feelings of guilt for being responsible for what happened to their children were also common[219]

More than 5 years have passed since the publication of this Clinical Practice Guideline, and its validity has not been reviewed

3	Significant association between the levels of parental satisfaction and items such as: suspicions of parents considered by professionals, the possibility to ask questions when they are given the diagnosis, the quality of the information received and professional treatment[218]
Qualitative Research	A high percentage of parents did not receive additional information or advice or support on the nature of ASD when they received the diagnosis. Many parents suggested that communication with professionals could be improved if they were suggested how to contact support groups or parent groups, and if there was a possibility to get in contact with Specialised Care professionals in an easier and more common way. Parents called for better training and guidance regarding ASD for professionals[220]
3	Between 71-73% of primary carers relied on media sources (Internet, books, videos, etc) for information, 42% on conferences / workshops and 42% on other parents. Only 15-20% resorted to health or educational professionals. 64% reported they belonged to a parents' association. Most professionals, 82% gave some additional information at the time of early diagnosis, compared to 18% who did not provide either additional information or referred to specialist services. 221

Recommendations

√	Practitioners should provide information to parents about what is appropriate child development
D	It is recommended to always address any concerns parents may have about the behaviour or development of their children to improve their satisfaction with the treatment received by the professional
Q	It should be remembered that new parents or parents without previous references may have more difficult to detect disorders in their children's development
√	When there is a suspicion of ASD, parents should be warned but not alarmed, making them aware of the importance of immediate evaluation
√	Professionals should provide correct information to help parents realize their child's problems, because some families have difficulty recognizing, understanding and accepting the disorder, especially when professionals are giving completely unexpected information
√	It is recommended NOT TO USE the terms "disorder" or "autism" (since a diagnosis has not been made yet), and use expressions such as: your child seems "not to have a communicative and social development appropriate for his or her age"
√	Action must be taken to the feelings of fear and denial of parents by being positive, non-judgmental and actively listening to concerns about the referral to Specialised Care
D	If there is any suspicion of ASD, one must be agile and provide information on the diagnostic process (referral), to increase satisfaction and confidence of parents and reduce their uncertainty (Appendix 8)

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√	The Health Care Team, especially the social worker, should take an "enabling" role when providing and interpreting information together with families

8.2 Aspects to consider when providing information to parents after a diagnosis of ASD

After a diagnosis of ASD, the family undergoes a very painful process that experts compare to the grief process, in this case mourning the loss of a "normal child." The stages of grief pass through: shock and initial denial, anger and resentment, depression and acceptance[38].

One of the first realities parents have to face is that children with ASD have an unchanged external appearance. Therefore, those ASDs such as Asperger disorder or autism not associated with intellectual disabilities can create more confusion to parents, as they have received a diagnosis difficult to understand when their children are apparently normal or intelligent. This is also due to ignorance and the stereotypes existing on these disorders.

Raising a child with ASD is a challenge for parents, which, upon acceptance of the diagnosis, should begin to develop adaptive strategies that promote the development of their children and the family as a whole. As they acquire the control of the situation, the interaction with their children will be better and this will be reflected in mental and physical health of parents[222].

In this process, we must not forget the feeling of isolation that is felt among caregivers of children with ASD, especially the one who takes the role of primary caregiver, mostly mothers. At a social level, there is little knowledge of these disorders so parents experience the stigma from the rest of society about their children. Such isolation can make the primary caregiver avoid any social interaction to avoid the stigma[9].

It is also frequent that the primary caregiver, the mother most of the time, has higher levels of stress and burnout for acquired responsibility[223], which exposes her to be blamed or criticised when there are problems with regard to the upbringing of the child[7].

In any case, one must be positive in the messages that are transmitted, as with time and the necessary support, the family will develop appropriate and functional coping strategies. Raising a child with ASD or with a disability in general, also provides important and enriching aspects at a personal and family level, as described by the families[224].

Qualitative Research

A phenomenological qualitative study explored the research process of raising a child with autism and presented the following findings[219]:

- Initial feelings after a diagnosis of ASD are similar to the stages of grief, in this case mourning the loss of a "normal child", going through the initial shock, denial, anger and resentment, depression and acceptance.
- The stress of caring for a disabled child is going to affect different components and family relationships: spouse, siblings and others.
- The participants expressed the tendency for one of the carers to assume the role of primary caregiver. The sense of responsibility was associated with increased stress.

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A descriptive, qualitative and quantitative study got information about parents' reactions to a diagnosis of autism through self-administered questionnaires (109 families responded) and focus groups of parents and professionals. Among the main findings it concluded that[38]:

Qualitative Research, Descriptive Study (3)

- Many parents had mixed feelings about the diagnosis received, as although it was a relief to have a diagnosis and understand the strange behaviour of his or her son, it also increased concerns about the future of their children.
- Upon acceptance of diagnosis and feelings of anger, fatigue, frustration and calm can alternate. They also express ignorance about ASD due to society and even some professionals.

In a descriptive study, 63 mothers of different social and cultural contexts were interviewed to explore the reactions, memories and beliefs about diagnosis and emotional health status of mothers.

Descriptive Study (3)

The acceptance process of the diagnosis is perceived as a process similar to the stages of grief. Among the main conclusions it describes how when there is acceptance of the diagnosis, mothers regain control of the situation, begin to interact optimally with their children and this results in a better state of mind for the mothers[225].

A descriptive study which evaluated the stress levels of 174 mothers and 43 fathers of children with ASD concluded[222]:

Descriptive Study (3)

- The stress level is inversely associated with the coping capacity of parents. Parents who chose avoidance or denial strategies had higher stress levels
- The stress level is directly related to the severity of symptoms and behavioural disturbances of the children, and inversely associated with social supports, family and therapeutic received.

A pre-post intervention study assessed the levels of stress and adaptability in 37 parents of children with ASD in comparison to 23 parents of children with normal development before and after being included in a children's program. The most relevant results are[226]:

Pre-Post Study (1-)

- Parents of children with ASD had higher levels of stress (59% of mothers and 35% of fathers) compared to parents of children without ASD (17% of mothers and 13% of fathers) before the program.
- The level of stress of mothers of children with ASD decreases significantly (from 59% to 46%) when the children access therapy programs, although this reduction did not occur with the fathers.

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A qualitative study described the coping capacity of parents of children with autism not associated with intellectual disability or Asperger disorder by introducing the gender perspective. In total 32 mothers and 21 fathers of 33 families from Brisbane, Australia were interviewed. The main findings are[227]:

Qualitative Research

- The role of the primary caregiver rests mainly on mothers, something that can be perceived in the levels of stress. The reduction of stress in mothers also affects the fathers. In addition, many of the mothers had to leave their jobs, as their children with ASD require more care, which implies full dedication to the care of the household and children.
- Differences between the impact of childcare on fathers and mothers could be observed. In contrast, the fathers argue that what is most striking in their lives comes indirectly from the impact of stress from their wives, rather than the impact of the disability of their children in itself.

A case-control study assessed parental depression in 216 families of children with autism and / or intellectual disabilities (ID) versus a control group of families of children without autism or ID using the Beck Depression Inventory (BDI) scale. The main outcomes were[228]:

Cases and control (2+)

The mothers of children with autism have higher levels of depression than mothers of children with ID without autism, which in turn have higher levels of depression than: fathers of children with autism, fathers of children with ID without autism, and parents from the control group.

A descriptive study carried out in Spain evaluated the burnoutl of the main carer of children with ASD (Zarit scale) as well as the physical (SF-36) and mental (SCL-90) health status. The sample was performed on 40 primary caregivers (38 women and 2 men). The main results were[7]:

Descriptive Study (3)

- High burnout in caregivers, reaching 72.5% in the range of intense burden.
- With regard to mental health, 57.5% is above the suffering average of the general population.
- In the physical dimension the results are indicative of poorer health, higher rates above the 50th percentile in physical pain (95%), vitality (87.5%), social function (85%) and mental health (82.5 %).

The Child Health Survey of New York, from 2003, assesses 61772 children (0-17) of which 364 have a diagnosis of ASD. Within the analysed data, it assesses mental health status and coping of mothers of children with autism. After the analysis, it can be observed that mothers of children with ASD have higher levels of stress and are more prone to mental and emotional problems that the mothers of the general population, even after being adjusted according to the socio-demographic level and the child's social skills[229].

Descriptive Study (3)

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Another descriptive study involving 62 families of children with ASD through a self-administered questionnaire explored the beliefs of parents about ASD and their implications for health care professionals. The most relevant results are[217]:

Descriptive Study (3)

- Most parents cited vaccines, genetic predisposition and environmental exposure of the mother during pregnancy as causes of ASD. Despite having received information about the causes, 26% still believed in the immunisation as probable cause and would have delayed or refused such immunisation.
- The average age at which parents noticed developmental delays or alterations were 9 months old, with an average age of diagnosis of 2 years and 3 months.
- Most parents (87%) would have resorted to the use of alternative or complementary medicine (without tested evidence) for their children, with an average of six different treatments used.
- Parents who perceived a long delay in the diagnosis or who had tried different therapies, tended to rely less on health professionals, but without statistical significance ($p = 0.2$ and $p = 0.07$ respectively).

A primarily qualitative mixed study which undertook a focus group and 15 semi-structured interviews with mothers of children with ASD explored the factors associated with the level of stress, depression and future expectations of their children. The main results are[39]:

Qualitative Research
Descriptive Study (3)

- Some mothers blamed themselves as the cause of ASD either due to the hereditary component or because they did not discard the idea of immunisation with the MMR vaccine as cause.
- Others however, were not concerned with the origin but for the future of their children.
- A large proportion felt responsible for the care of their children, which included feelings of frustration, isolation and anger at the lack of support services and for feeling overloaded.

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A mixed study analysed the dynamics and coping strategies of 52 parents of children with ASD, using qualitative methodology (semi-structured interviews) and quantitative with the use of adaptability and cohesion scales (FACES III), personal evaluation of family crisis (F-COPES) and perception of family support and friends (PSS-FA and PSS-FR). Some of the most relevant results of quantitative analysis were [224.230]:

Qualitative Research Descriptive Study (3)

Those families categorised as cohesive or involved developed effective coping strategies and managed stress better, regardless of the support received. There were no significant differences between the perceptions of parents, except that mothers received more assistance from other relatives and friends. No differences were found in the frequencies of different types of cohesion between this sample of families and standardised families.

As for the qualitative data, it is significant that for all the families it was very important to determine the causal origin of autism of their child. Moreover, the negative conceptualisation of "raising a child with autism" does not help in developing effective strategies. Almost all parents were able to describe positive learning experiences as a result of having a child with autism. They said they were more patient, compassionate, humble, and tolerant. Instead of seeing their situation as something negative, many parents see it as a positive adaptive process, which gives more meaning to their relationships with family and friends and has enriched them personally and makes the family stronger.

Other studies have addressed the adaptation process of the families throughout time. A longitudinal ethnographic study carried out during 10 years highlighted that the first years after diagnosis were very stressful for families, but with time, the perception of stress and stigma decreased. Parents attributed this improvement to an increased ability to manage and adapt to the child's behaviour and a perception of relatively high levels of satisfaction and services. However, in families with a child with aggressive or violent behaviour, emotional stress levels persist, with little help or intervention resources[231,232]

Qualitative Research

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Summary of evidence

Qualitative Research	The initial feelings after a diagnosis of ASD are similar to the stages of grief going through the initial shock, denial, anger and resentment, depression and acceptance. The stress of caring for a child with a disability will affect the different components and family relationships: spouse, siblings and others One of the caregivers assumes the role of primary caregiver, thus provoking the sense of responsibility to be associated with increased stress[219]
Qualitative Research 3	Many parents had mixed feelings about the diagnosis received, because, although it meant a relief to diagnose and understand the strange behaviour of their child, there was also a growing concern about the future of their children. Following the acceptance of the diagnosis and with time, the feelings of anger, fatigue, frustration and quiet can alternate. They also expressed ignorance about ASD by society and even some by professionals[38] When it comes to accepting the diagnosis, the mothers develop skills to retrieve the control of the situation, begin to interact optimally with their children and this results in a better state of mind in mothers[225]
3	Following the acceptance of the diagnosis and with time, the feelings of anger, fatigue, frustration and calm can alternate[38]
3	The stress level is inversely associated with the coping capacity of parents, the social, family and therapeutic support received, and directly related to the severity of the symptoms of the children, as well as with strategies of avoidance or denial of parents[222]
1-	Parents of children with ASD have higher levels of stress compared to parents of children without ASD. The stress level of mothers decreases when children enter treatment programs[226]
Qualitative Research	The role of primary caregiver rests largely on mothers, and this role is perceived with stress. Many mothers had to leave their jobs as their children with ASD require more care, which implies a full dedication to the household and children There are differences observed between the impact of childcare on fathers and mothers. What most impresses fathers are the stress levels of mothers[227]
2+	Mothers of children with autism have higher levels of depression than mothers of children with intellectual disability without autism, which in turn have higher levels of depression than: fathers of children with autism, fathers of children with ID without autism, and parents from the control group of families without autism or DI[228]
3	High burnout is observed in caregivers, reaching 72.5% in the range of burden. With respect to mental health 57.5 is above the suffering average of the general population. In the physical dimension the results are indicative of poorer health, being above the 50th percentile in physical pain (95%), vitality (87.5%), social function (85%) and mental health (82.5%)[7]
3	Mothers of children with ASD have higher levels of stress and are more prone to mental and emotional problems that the mothers of the general population, even after adjusting according to the socio-demographic level and the child's social skills[229]

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3	26% of parents still believed in immunisation as the probable cause and would delay or refuse such immunisations according to their experience The average age at which parents noticed developmental delays or alterations were 9 months old, with an average age of diagnosis of 2 years and 3 months Most parents (87%) had resorted to the use of alternative or complementary medicine without evidence tested for their children, with an average of six different treatments used There is a trend in the data on parents trying different therapies, they tended to rely less on health care professionals (p = 0.07)[217]
Qualitative Research 3	Some mothers blamed themselves as the cause of ASD and were concerned about the future of their children. A large proportion felt responsible for the care of their children, feeling frustration, isolation and anger at the lack of support from social services as well as being overloaded[39]
Qualitative Research 3	Families that involve themselves more and are more united developed effective coping strategies. To determine the causal origin of autism of their child was very important for all the families. The negative conception of "raising a child with autism" does not help to develop effective strategies. Almost all parents were able to describe positive learning experiences as a result of having a child with autism[224.230].
Qualitative Research	The well-being perception of families is greater as time passes and it increases treatment strategies, although in families of children with severe behaviour disorders, emotional stress levels remain high[231.232].

Recommendations

G	Professionals must accept and understand the first emotions resulting from early diagnosis as part of the process of accepting the new reality
D	Professionals should provide objective and simple information to parents about what are ASDs, their etiology, and respond to any questions that may apply, so that they can control the situation as soon as possible and increase their ability to cope with it (Appendix 9)
D	Professionals should be wary of burnout, depression and stress phenomena of the primary caregiver, usually the mother
Q	Professionals should be positive in the messages that are transmitted so that the process of adaptation and acceptance of families is effective, since they must rebuild their social conceptualisation of what it means to have a child with autism, as well as acquire management skills
√	The Health Care Team, especially the social worker, should take an "enabling" role when providing and interpreting information together with families

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9. Strategies for monitoring children with ASD in Primary Care

The questions that are going to be answered in this chapter are:

- How to plan the visit of children with ASD in Primary Care?
- What issues should be explored / monitored in the Primary Care visit of children with ASD?

Once the child has a confirmed diagnosis of ASD, Primary Care professionals should continue to monitor the development to prevent any complications that may arise and continue with preventive care and encouragement of the child's health (control of growth, vaccinations, fostering of healthy habits such as exercise, and others) and resolve any other health problems associated or not with autism.

Dealing with a child with ASD at the surgery can be complex and requires the development of certain strategies in coordination with the family so as to be properly assessed, taking into account the special characteristics of children with ASD in their limited communication skills, and behaviour disorders such as intolerance to changes in their routines, hypersensitivity to scans, and others. These characteristics may interfere in the diet, sleep and other problems as certain behaviours can lead to risks involving not only the child's health but also its physical integrity[6, 36, 46, 136,148,233,234].

According to experts, children with ASD can have any of the following behavioural disorders[6, 36, and 46,136,148,233-235]:

CPG
Expert opinion (4)

- They tend to reject noisy, strange, crowded and untidy places.
- They generally do not tolerate changes in their daily routines.
- In some cases, they can show rejection certain to procedures because they involve physical contact.
- As far as dental hygiene, they may have an aversion to brushing their teeth, or can even eat toothpaste.
- They tend to have attachment to certain toys or develop their own restricted interests.
- Due to sensory impairment, different thresholds of perception with hyper or hypo-sensibility may arise, in a way that these can mask the pain.
- They can also perform dangerous or atypical behaviours such as wearing no warm clothes when it is cold or raining, tighten their clothes too much, eat food which is too hot or wash themselves with extremely hot water, inhale or take toxic substances as they might like their smell or taste. Sometimes they may provoke self-harm with injuries, bites, pinches, self-inflicted in times of stress.

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Some studies consider the IQ as a significant predictor of adaptive behaviour, as it is a limiting factor particularly in those children with intellectual disabilities (IQ below 80) [156,236].

Cohort studies (2+)

Children with ASD have impaired ability to communicate at different levels. This, combined with sensory disorders, increases the problems to express certain symptoms like pain or discomfort. Even those children with ASD who have capacity for language, suffer from linguistic disorders, and thus do not understand double meanings or jokes, as happens in the Asperger syndrome.

Descriptive study (3)

RS of different types of studies (1+)

According to a systematic review of the benefits of Alternative and Augmented Communication (AAC) and other series of case studies which use alternative means of communication to language, those interventions that provide visual support (such as the use of pictograms, photographs, cartoons, drawings, imitation) facilitate social communication processes[148,.237- 240]. However, there is not enough evidence to suggest that these interventions increase the production of language in children with ASD[241].

According to a systematic review of 31 articles that assessed effectiveness and unintended effects associated with the trivalent MMR vaccine (Measles, Mumps and Rubella), there is currently no evidence which can link vaccination with the development of ASD[126].

RS of different types of studies (1++)

The disturbance in sleep patterns is common in children with ASD (between 44%-83%). These conditions will inevitably affect the parent's own sleep patterns. There are a variety of treatments available from behavioural to pharmacological strategies[242-245].

Cases and control (2+)

A study of cases and controls could not find evidence that children with autism have specific gastrointestinal disorders[246]. However, there does exist evidence that behavioural disorders may be expressed as eating disorders, such as pica, or rejection of food for its taste, colour, texture or smell and can often lead to nutritional deficiencies. These disorders can also be approached from a psychobehavioural therapeutic standpoint[247-249]. Despite the absence of specific digestive problems, several studies confirm the existence of functional gastrointestinal problems (such as constipation and other bowel disorders, among others), that in this group of population can have major behavioural and adaptive consequences[250-252]

Cases and control (2+)

There is insufficient evidence to support the use of specific diets such as gluten-free diet and casein[127].

RS of ASDs (1++)

Nor is there sufficient evidence to support the use of secretin, dimethylglycine, vitamins, Omega-3 fatty acids and dietary supplements as a treatment of ASD[46,136,253-257].

ASD (1+) RS of ASDs (1++)

There is insufficient evidence to support the use of alternative therapies such as auditory integration training and other sound therapies including music modified by filtering and modulation, and the use of hyperbaric oxygen for ASDs [258.259].

RS of ASDs (1++)

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ASD (-1)
Clinical Cases (3)

There is insufficient evidence to support the use of immunotherapy, chelating agents and antifungal treatments of ASD[46,126,136,260,261]. Immunotherapy treatments are not supported by quality clinical trials and in the case of anti-fungals, no tests have been performed. Nor is there evidence to support the chelating agent therapy. On the other hand, no autism causal relationship has been found between the vaccines, which used mercurial derivative (thimerosal) as a preservative 126. Furthermore, these treatments are potentially dangerous[261-264]

Summary of evidence

3	Children with ASD may have risk behaviours (self-injury), sleep disorders, eating or hygiene disorders, sensitivity to changes and hide certain symptoms like pain, which can lead to stress or anxiety when attending the physician's office[46,136,138,148,235]
2+	Some studies considered the intellectual quotient (IQ) as a significant predictor of adaptive behaviour, being a limiting factor especially in those children with intellectual disabilities (IQ below 80)[156,236]
3 1+	Alternative support language interventions such as visual language improve the communication processes in children with ASD[148,237,240,241]
1++	No relationship between vaccine immunisation and ASD development has been proven[126]
2+	Children with ASD often have sleep disorders[242-245]
2+	There is no evidence to support that children with ASD have specific gastrointestinal problems compared with children without ASD, but may have eating disorders such as pica, or rejection of certain foods, and functional gastrointestinal problems in this population group may have greater adaptive and behavioural consequences[246-252]
1++ 1+	There is no evidence to support the use of gluten-free diet and casein and alternative treatments such as secretin, vitamins B6 combined with magnesium, Omega-3 fatty acid, dimethylglycine, hyperbaric oxygen or auditory integration training[127136253-259]
1- 3	There is no evidence to support the use of antifungals, chelating agents and immunotherapy[46,126,136,260-263]

Recommendations

√	Professionals should take account of the symptoms and behavioural disorders of children with ASD to prepare, in coordination with parents, the visits of the child to prevent overstimulation and confusion or potential health problems or risk behaviours
C	It would be useful to register children with ASD by maturation level (IQ or developmental level) as it is a key prognostic factor, which determines the global evolution (learning, communication, social skills)

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B	Professionals can use visual support interventions such as the use of pictograms, cartoons, dolls imitation, to support communication processes in children with ASD. Example: to show them the instruments, procedures and techniques, exploration of pain and other symptoms
D	Professionals should use short simple sentences, without double meanings, to communicate previously to the child what he or she will do, and use direct orders when asking them for something, using the support of parents to understand and communicate
√	In the visit of children with ASD, Primary Care professionals should: – Coordinate with the parents so that they anticipate the visit to the child – Try not to wait for a long time in the waiting room, especially if it is full of other patients – Trust in the management criteria of parents – Welcome the child in a calm and orderly environment – Reinforcements with prizes (toys, stickers and others) may facilitate future visits – In case a certain procedure that involves physical contact is required, it is necessary to assess risk/ benefit of doing so. For example, a blood test, for which the child should be as much prepared as possible, accepting more flexible rules such as allowing the parent to accompany the child or adapting the procedure if necessary – Be aware of disorders and behavioural sensitivity to detect: masked symptoms such as pain, certain harmful or dangerous habits like inhaling or ingesting toxic substances, self-harm – Adjust the format of the medication to the child's preferences as much as possible (i.e. syrups instead of tablets)
D	Professionals should monitor nutritional status of children with ASD, recommending dietary supplements if necessary or even referring
D	Professionals should manage the gastrointestinal problems of children with ASD in the same way as those of children without ASD, taking into account that the existence of functional gastrointestinal problems (constipation, bowel disorders, and others) can have major adaptive and behavioural consequences than in population without ASD
√	Professionals should inform parents about healthy habits such as diet or exercise, leisure and free time activities, selecting those that, within the limits of the family and those provided by the community, provide enjoyment and child welfare
D	Monitor oral health of children with ASD. In case of eating toothpaste, do recommend one without fluoride. Patients and parents' associations and parents can provide guidance on which dentists have experience attending children with autism, as sometimes certain changes in the procedures may be necessary. The same applies if ophthalmic check-ups are required

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B	All children with ASD should follow the immunisation schedule established just like any other children, including vaccination of MMR (rubella, measles and mumps). Professionals should inform about the importance of this to parents
C	Professionals should monitor the presence of sleep disorders, using as a first step behavioural strategies to manage them and training parents on this issue
C	Professionals should inform parents that there is no evidence that ASDs are associated with digestive disorders
A,B	Professionals should inform parents that currently there is no evidence of the effectiveness of diets free of gluten and casein, secretin, vitamin B6 plus magnesium, Omega-3 fatty acid, dimethylglycine, hyperbaric oxygen therapy and music therapy for specific treatment of ASD
C,D	Professionals should inform parents that currently there is no evidence of the effectiveness of antifungal therapy, chelating agents and immunotherapy
√	Professionals should inform parents that there are currently some "alternative treatments" that have no scientific basis to support their use and could be potentially dangerous, so they should be advised to consider only of those interventions recommended by professionals

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10. Dissemination and implementation

The questions that are going to be answered in this chapter are:

- What is the guide's dissemination and implementation strategy?
- What are the indicators for monitoring the key recommendations of the guide?

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10.1. Dissemination and implementation strategy

Clinical practice guidelines are useful to improve the quality of the care provided and the outcomes of patients. The big challenge now is to get the adhesion of professionals. An implementation strategy aimed at overcoming the barriers in the environment in which they are to be applied is required.

The plan to implement the guidance on the detection and approach to children with autism spectrum disorders in primary care includes the following interventions:

- Presentation of the guide to the media by the health authorities.
- Presentation of the guide to the Primary Care and Special Care management and submanagement branches of the various Regional Health Services.
- Institutional presentation of the guide in collaboration with the Quality Agency of the Ministry of Health and Social Policy to the various scientific and professional societies involved.
- All presentations will highlight the informative material produced for the patient in order to facilitate its distribution among all health professionals as well as to the parents of children with this problem.
- The informative material for parents will be presented to the parents' associations so that in turn they can also distribute and disseminate it.
- Effective and targeted distribution to professional groups involved (paediatricians, nurses, family doctors and primary care social workers) to facilitate dissemination.
- Interactive presentation of the guide in health centres by local opinion leaders.
- Dissemination of the guide in electronic form on the website of the Ministry of Health and Social Policy, of GuíaSalud, of the UETS and of scientific societies and associations involved in the project.
- Publication of the guide in scientific medical journals.
- Establishment of criteria for good care of patients with ASD in the programme contracts and contracts for clinical management, as set out in the guide
- Evaluating effectiveness of implantation, establishing systems of clinical decision sup-

port, integrating the guide and the indicators selected in the software used in Primary Care.

10.2. Indicators proposed

The authors of this CPG have designed a series of indicators that should be measurable through the information system in Primary Care in order to evaluate both health care to the patient with ASD and the potential impact of the implementation of the guide. It was not the purpose of the authors to design a comprehensive and detailed evaluation involving the use of all proposed indicators. It is intended to provide a tool for clinicians and managers concerned, which may be useful to design specific assessment of the care provided to children with ASD in Primary Care.

Two types of indicators are proposed:

- Monitoring indicators: This set of indicators is aimed to monitor the distribution of patients by using procedures and assessment tools suggested in the guide.
- Performance indicators: Based on the recommendations proposed in this guide and therefore the available scientific evidence and consensus of professionals. Although the proposed performance standards should be 100%, the reality of the context of Primary Care was taken into account when establishing these standards.

Monitoring indicators

Numerator: Number of children with suspected ASD that attend a healthy child consultation

Denominator: Number of children attending a healthy child consultation *

* Healthy child consultation means the compliance of the visits to consultation according to the program agenda established for any healthy child in every health care area

Numerator: Number of children diagnosed with ASD that attend a healthy child consultation

Denominator: Number of children attending a healthy child consultation *

Numerator: Number of children with suspected ASD referred to Early Care

Denominator: Number of children with suspected ASD referred to Specialised Care **

** Specialised Care is defined as referral to Early Intervention units, local referral units and professional clinicians

Numerator: Number of children with ASD, monitored in the healthy child consultation, which are referred to Specialised Care due to associated comorbidity

Denominator: Number of children with ASD who are monitored in the healthy child consultation

Performance indicators

Numerator: Number of Children to which development is assessed at least between 8-12

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months, 2-3 years and 4-5 years

Denominator: Number of children assessed in the healthy child consultation
Standard: 100%

Numerator: Number of children assessed with the Haizea Llevant Development Scale

Denominator: Number of children assessed in the healthy child consultation
Standard:> 90%

Numerator: Number of children between 16 -30 months of age with suspected ASD evaluated with the M-CHAT scale

Denominator: Number of children between 16 -30 months of age with suspected ASD
Standard: 100%

Numerator: Number of children older than 5 years of age with suspected ASD evaluated with the Autonomous Scale

Denominator: Number of children older than 5 years of age with suspected ASD
Standard: 100%

Numerator: Number of children under 16 months of age and between 30 months and 5 years with suspected ASD referred to Specialised Care

Denominator: Number of children under 16 months of age and between 30 months and 5 years with suspected ASD
Standard: 100%

Numerator: Number of children with positive M-CHAT or Autonomous Scale with altered result referred to Specialised Care

Denominator: Number of children with positive M-CHAT or altered Autonomous Scale assessed in the healthy child consultation
Standard: 100%

Numerator: Number of siblings of children with ASD to be watched for signs of ASD in the healthy child consultation

Denominator: Number of siblings of children with ASD attending a healthy child consultation.
Standard: 100%

Numerator: Mothers or fathers of children with suspected ASD who are provided with written information

Denominator: Mothers or fathers of children with suspected ASD evaluated in the healthy child consultation
Standard: 100%

Numerator: Mothers or fathers of children with ASD who are provided with written information

Denominator: Mothers or fathers with children with ASD attending a healthy child consultation
Standard: 100%

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11. Recommendations for future research

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In the studies assessed for the development of this guide, in general, a low level of evidence has been observed, as the methodological approaches are primarily descriptive, with very few analytical studies. For this reason, it is urgent not only to establish more research lines on ASD but also to increase the number of analytical studies and trials.

Depending on the scope and objectives of our guide, the basic research priorities should include:

- Studies on environmental etiological factors (heavy metals, organic compounds and others) to enable preventive measures, as well as the expansion of research into the genetics of autism (family patterns) towards a more comprehensive genetic counselling.
- Study of the most frequent comorbid disorders and complications associated with autism for proper tracking and monitoring in Primary Care and in Specialised Care.
- Studies that combine behavioural, neuropsychological and developmental variables, to develop the diagnostic criteria laid down in the ICD-10 and DSM-IV manuals and more specifically to differentiate subgroups and subtypes in the spectrum.
- Studies on analysis of initiation and early evolution of the disorder patterns to establish the feasibility minimum age of diagnosis.
- Studies on the evolution of somatosensory, motor, communicative and social development of children with ASD.
- Studies to support early detection, in terms of demonstrating better health outcomes of interventions depending on the intervention age of onset.
- Development and validation of ADS screening instruments that meet the criteria for group screening.
- Studies on the quality of life of children with ASD and their families.
- Studies on the primary caregiver's burden of children with ASD.
- Studies with qualitative methodology which inform about the experiences of parents and families of children with ASD at early stage screening and referral to Specialised Care, use and satisfaction of health services in Primary Care following diagnosis; and the experiences of professionals from both Primary and Specialised Care to identify gaps in knowledge, resources and problems to care for children with ASD and their families.
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12. Appendices

Appendix 1. Levels of evidence and grades of recommendation

Levels of evidence and grades of recommendation (SIGN)[265]

Levels of scientific evidence

1++	High-quality meta-analysis, systematic reviews of clinical trials or high-quality clinical trials with very low risk of bias.
1+	Well-conducted meta-analysis, systematic reviews of clinical trials or well-conducted clinical trials with little risk of bias.
1-	Meta-analysis, systematic reviews of clinical trials or clinical trials with high risk of bias.
2++	High quality systematic reviews of cohort studies or case-control study. Cohort or case-control studies with very low risk of bias and with high probability to establish a causal relationship.
2+	Cohort or well conducted case-control studies with low risk of bias and a moderate probability of establishing a causal relationship.
2-	Cohort or well conducted case-control studies with low risk of bias and a moderate probability of establishing a causal relationship.
3	Analytical studies, such as case reports and case series.
4	Expert opinion.

NOTE: Studies on qualitative methodology "Qualitative Research." This category is not covered by SIGN. The studies included were evaluated regarding methodology, and they included in this category those studies, which were more rigorous.

Grades of recommendation

A	At least one meta-analysis, a systematic review or a clinical trial rated as 1 ++ and directly applicable to the target population of the guide; or a volume of scientific evidence consisting of studies rated as 1+ and with great consistency among them.
B	A volume of scientific evidence consisting of studies rated as 2++, directly applicable to the target population of the guide and showing great consistency among them; or extrapolated evidence from studies rated as 1 ++ or 1 +.

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C	d A volume of scientific evidence consisting of studies rated as 2 +, directly applicable to the target population of the guide and showing great consistency among them, or extrapolated evidence from studies rated as 2 + +
D	Level 3 or 4 scientific evidence, or extrapolated evidence from studies rated as 2 +

Studies classified as 1- and 2- should not be used in the process of developing recommendations due to their high potential

√ [1]	Recommnded practice based on clinical experience and consensus of the development team.

1. Sometimes the development group realises that there is some important practical aspect, which should be emphasised, and for which there is probably no scientific evidence that supports it. In general, these cases are related to some aspect of treatment considered good clinical practice and usually no one would argue about them. These aspects are rated as points of good clinical practice. These messages are not an alternative to the recommendations based on scientific evidence but should be considered only when there is no other way to highlight this aspect.

Q	Evidence from studies with quality and relevant qualitative methodology. This category is not covered by SIGN.

Levels of evidence and grades of recommendation for diagnostic studies (NICE adaptation of levels of evidence of the Oxford Centre for Evidence-based Medicine and the Centre for Reviews and Dissemination)[266]

Levels of scientific evidence	**Type of evidence**
Ia	Systematic review with homogeneity of **level 1** studies.
Ib	**Level 1** studies.
II	**Level 2** studies. Systematic review of **level 2** studies.
III	**Level 3** studies. Systematic review of **Level 3** studies.
IV	Consensus, expert opinion without explicit critical appraisal.
Level 1 Studies	Meet: - Masked comparison with a valid reference test (gold standard). - Adequate spectrum of patients.

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Level 2 Studies	They have only one of these biases: - Non representative population (the sample does not reflect the population where the test will be applied). - Comparison with the inadequate reference standard (gold standard), (the test will be evaluated as part of the gold standard or the test result affects the implementation of the gold standard). - Comparison is not masked. - Case-control studies.
Level 3 Studie	Include two or more of the criteria described in level 2 studies

Recommendation	Evidence
A	Ia or Ib
B	II
C	III
D	IV

Appendix 2. NANDA diagnoses (North American Nursing Diagnosis Association) proposed for children with ASD and their relatives[144,145]

The standardised nursing care plan shown is aimed to the care of children from 0-6 years with Autism Spectrum Disorders (ASD) in the field of Primary Care. The problems which are more frequent in these children and their families are presented. For its description, we have used the labels of the NANDA taxonomy, including the nursing intervention codes (NIC) and the nursing outcomes (NOC).

Nurses from Primary Care and Specialised Care have participated, so as to reach a consensus, which ensures the continuity of the care provided and the coordination between the different care levels.

Finally, it must be remembered that standardised care plans involve an abstraction and should always be individualised and contextualised. That is, be understood as a proposal that each nurse must tailor to each child and family.

Diagnoses oriented to child with ASD

00 111 Delayed growth and development: Deviations from the rules for an age group

00 112 Risk of failure to develop: Risk of suffering a 25% or more delay in one or more areas of social or self-regulatory behaviour, cognitive, language or gross or fine motor skills

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NOC		NIC	
0120	Child development: 1 month	8274	Promoting development: child
0100	Child development: 2 months	5605	Education: infant stimulation
0101	Child development: 4 months	5566	Paternal education: family upbringing of children
0102	Child Development: 6 months		
0103	Child development: 12 months	8250	Care Development
0104	Child development: 2 years	4360	Behaviour Modification
0105	Child Development: 3 years		
0106	Child development: 4 years		
0107	Child Development: preschool		
0108	Child development: second childhood		

00 051 Impairment of verbal communication: Decrease, delay or lack of ability to receive, process, transmit and use a system of symbols

NOC		NIC	
0902	Communication	4976	Improving communication: speech deficits
0903	Expressive communication	5540	Enhancing the provision of learning
0904	Receptive communication	4720	Cognitive Stimulation
		5460	Contact

00 052 Impairment of Social Interaction: Ineffective or qualitatively insufficient or excessive social exchange.

NOC		NIC	
0116	Participation in games	5100	Enhancing socialisation
1502	Social interaction skills	4362	Behaviour Modification: Social skills
0915	Hyperactivity Level	4430	Therapy with games
		4352	Management of behaviour: hyperactivity / lack of attention

Family-oriented diagnoses of a child with ASD

00 126 Deficient knowledge (specify): Lack or deficiency of cognitive information related to a specific topic

NOC		NIC	
1803	Knowledge: disease process	7400	Health System guides
1824	Knowledge: medical care	5510	Health education
1806	Knowledge: health resources	5568	Parental education

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1823	Knowledge: health promotion	5632	Education: young child safety
1801	Knowledge: child safety	5630	Education: young child nutrition
1700	Beliefs about health	5520	Facilitating learning
1703	Beliefs about health: perception of resources	6610	Hazard identification

00 075 Provision to improve family coping: Effective management of the adaptive tasks of the family member involved in the challenge of the patient's health, which now shows need and willingness to improve his or her own health and development and that of the patient

NOC		NIC	
2204	Relationship between the primary caregiver and the patient	7140	Family support
2602	Family functioning	5240	Counselling
2605	Family involvement in professional health care	7280	Siblings support

00 073 Disabling family coping: Behaviour of a significant person which implies the disabling of his or her own capabilities and those of the patient to actually leave essential tasks so that both can adapt to the health challenge

NOC		NIC	
2506	Primary caregiver's emotional health	7110	Promoting family involvement
2508	Primary Caregiver's Welfare	7120	Family mobilisation
2208	Family Caregiver's Stressors	4700	Cognitive restructuring
2210	Possible resistance to family care	5400	Enhancing self-esteem
1606	Participation in decisions about health care		

00 074 Family coping involved: The person who usually provides the main support in this case provides help, comfort, assistance or insufficient or ineffective encouragement, which may be necessary for the patient to manage and dominate those adaptive tasks related to his or her health

NOC		NIC	
2600	Overcoming family problems	5270	Emotional Support
2604	Standardisation of the family	7040	Primary Caregiver Support
2205	Performance of primary caregiver: direct care	5250	Support for decision making
2206	Performance of primary caregiver: indirect care	5230	Increase coping skills
		7100	Stimulation of family integrity
		7200	Foster family standardisation
		5520	Facilitating learning

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00 164 Provision to improve parental role: Pattern of providing an environment for children or other dependents that is sufficient to promote growth and development and can be strengthened.

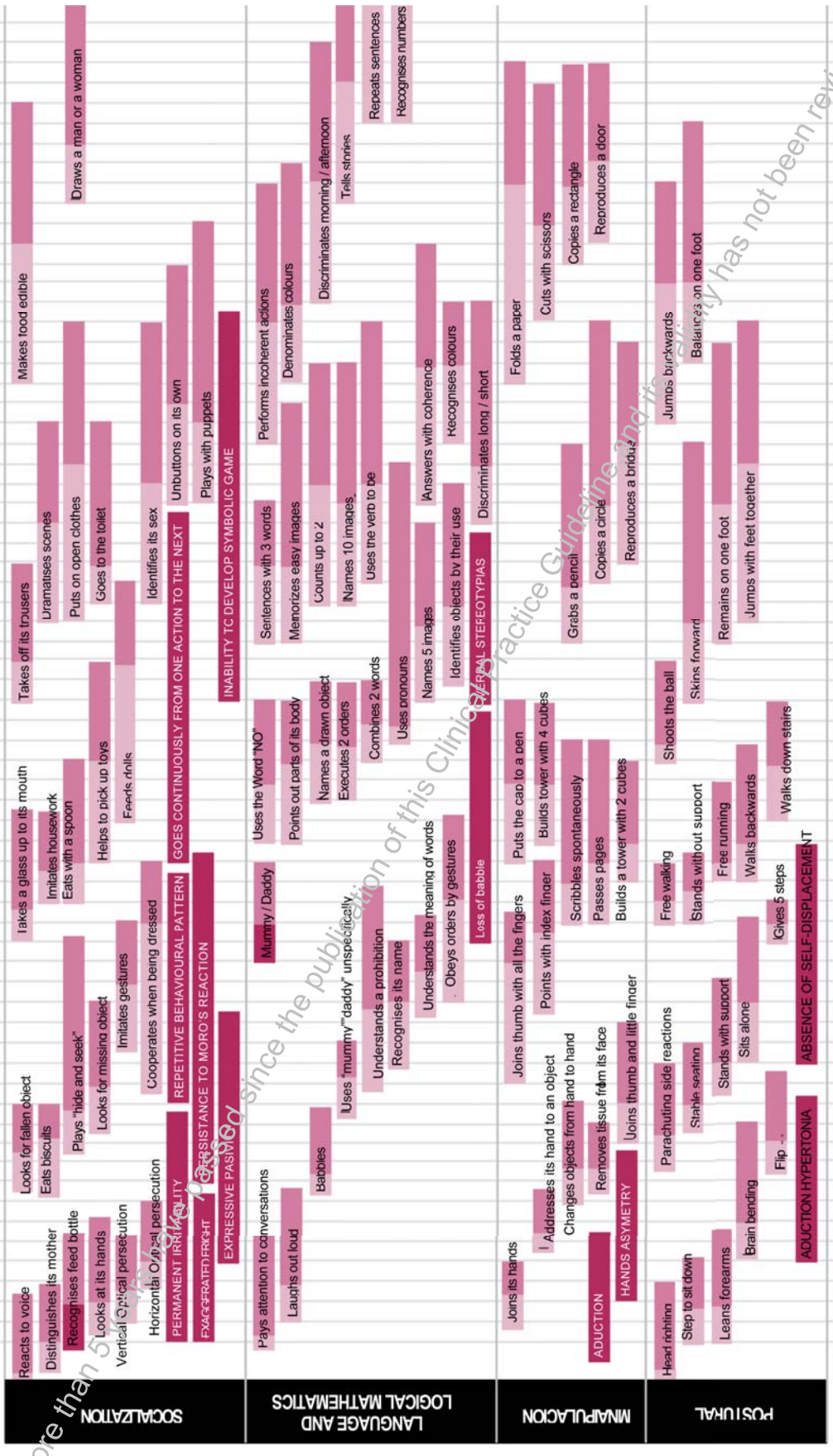
NOC		NIC	
2211	Parenting	8300	Promoting parenthood
1603	Behavioural Health Search	5566	Paternal education: family upbringing of children
1602	Conduct health promotion	5568	Paternal education: child

00 058 Risk of deterioration in the relationship between parents and the infant / child: Alteration of the interactive process between the parents or a significant person and the infant / child that fosters the development of a reciprocal protective and training relationship.

NOC		NIC	
1500	Parent-child bonds	6481	Environmental management: Coming together process
2211	Implementation of the parenting role	6710	Fostering the bringing together
1704	Beliefs about health: perception of threat	5370	Empowerment roles
		5480	Clarification of values
		4480	Facilitating self-reliance

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Appendix 3. Haizea- Llevant development Monitoring Table



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Interpretation Rules of the Haizea-Llevant development table:

The table is divided into four key areas: sociability, language, handling and postural with different tests performed for each of them.

The tests are represented in the table in the form of horizontal bars with dark and pale pink colour indicating the percentage of children who perform an action.

The beginning of the bar (pale pink) indicates 50% of children; the change of colour (Medium Pink) indicates a 75% and the end of it at this age indicates that 95% of children already do the action studied.

We also find in the table some warning signs represented as lines of dark pink .

The correct order of the evaluation of the table is "reading it" from left to right and from top to bottom.

Draw a vertical line that corresponds to the age in months of the child (adjusted for less than 18 months of age in cases of prematurity).

Ask the family and check, if necessary, whether the child performs:

a) Those elements located to the left of the cut line.

b) Those elements that cross that line.

The examiner will assess the lack of acquisition of these elements, in some or all the areas, as well as the presence of warning signs to determine the need for further diagnostic studies.

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Appendix 4. Warning Signs

IMMEDIATE WARNING SIGNS[172]
– Does not babble, no gestures (pointing, waving good-bye) at 12 months.
– Does not say single words at 18 months.
– Does not say two-word spontaneous phrases (not echolalic) at 24 months.
– Any loss of language or social skills at any level at any age.
– Any loss of language skills or social level at any age.
– Any loss of language skills or social level at any age.

TABLE 6: SUMMARY OF ASD WARNING SIGNS BY AGE PERIODS
Note: The signs established in previous stages can appear at any age. These factors alone do not indicate ASD. They should warn professionals on the risk of ASD, so that the referral depends on the situation as a whole. They cannot be interpreted from the evolutionary point of view of ASD either, but as potential signs that can be found in children evaluated at the ages referenced.
Before 12 months
– Low frequency of use of the gaze directed at people
– Shows no advance when it is going to be held
– Lack of interest in simple interactive games like "peek-a-boo" or " give and take "
– Lack of social smile
– Lack of anxiety to strangers when about nine months
After 12 months
– Less eye contact
– Does not respond to name
– Does not point to "ask for anything" (proto-imperative)
– Does not show objects
– *Unusual* response to auditory stimuli
– Lack of interest in simple interactive games like "peek-a-boo" or " give and take "
– Does not look where others point out
– Lack of spontaneous imitation
– No social / communicative babbling as if it talked with adults
Between 18-24 months

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– Does not point the finger to "share an interest" (protodeclarative)

– Has difficulty to follow the gaze of an adult

– Does not to look where others point out

– Delayed development of understanding and / or expressive language

– Lack of functional play with toys or presence of repetitive forms of play with objects (i.e., put in line, open and close, switch on and off, etc)

– Lack of symbolic play

– Lack of interest in other children or siblings

– Does not usually show objects

– Does not respond when called

– Does not imitate or repeat gestures or actions that others do (i.e., grimaces, claps)

– Few expressions to share positive affect

– Used words before but not anymore (language regression)

From 36 months

Communication

– Absence or language delay or deficit in language development is not balanced by other modes of communication

– Stereotyped or repetitive use of language such as echolalia or refers to himself or herself in 2nd or 3rd person

– Abnormal intonation

– Poor response to its name

– Deficits in nonverbal communication (i.e., no pointing out and difficulty identifying and sharing a "focus" with the look)

– Failure of social smile to share enjoyment and respond to the smiles of others

– Achieves things by himself or herself, without asking

– Used words before but not anymore

– No varied representation games or social imitation appropriate to developmental level

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Social unrest – Limited imitation (i.e., clapping) or absence of actions with toys or other objects – Does not "show" objects to others – Lack of interest or strange approaches to children of its age – Little recognition or response to happiness or sadness of others – Does not join others in shared imagination games – Failure to initiate simple games with others or participate in simple social games – Preference for solitary activities – Strange relations with adults from an excessive intensity over to a striking indifference – Lack of social gaze
Disorders of interest, activities and behaviours – Insistence on routines and / or resistance to changes in unstructured situations – Repetitive games with toys (i.e., aligning objects, turning lights on and off, etc) – Unusual attachment to a toy or object which it always carries that interferes with his or her daily life – Hypersensitivity to sounds, touch and certain textures – Unusual response to pain – Unusual response to sensory stimuli (auditory, olfactory, visual, tactile and taste) – Strange postural patterns such as tiptoe – Motor stereotypy or mannerisms
Screening from 5 years *
Communication Disorders – Poor language development, including muteness, odd or inappropriate intonation, echolalia, unusual vocabulary for its age or social group – In the case where there are no deficiencies in language development, there is limited use of language to communicate spontaneously and a tendency to speak only about specific topics of its own interest (fluid language but hardly appropriate to the context)
Social unrest – Difficulty joining the game of other children or inappropriate attempts to play togetherLimited ability to appreciate cultural norms (in dress, style of speech, interests, etc.) – Social stimuli will produce confusion or displeasure – Inappropriate relationships with adults (too intense or nonexistent) – Shows extreme reactions to the invasion of its personal or mental space (intense resistance when to much pressure is put with different instructions to its focus of interest)

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Limitation of interests, activities and behaviours – Lack of flexibility and cooperative imaginative play, but usually creates alone certain imaginary scenarios (copied from videos or cartoons) – Difficulties of organisation in unstructured spaces – Inability to deal with changes or unstructured situations, including those which the children enjoy such as school excursions, when the teacher is absent, etc. – Collects data on certain topics of interest in a restrictive and stereotyped way
Other features – Unusual abilities and weaknesses profile (i.e., poorly developed social and motor skills, gross motor clumsiness) – General knowledge, reading or vocabulary may be beyond the chronological or mental age – Any significant history of loss of skills – Certain areas of knowledge may be particularly strong, showing amazing skills in areas such as mathematics, mechanics, music, painting, sculpture
* The warning signs from 5 years of age are more focused on those cases that may have been overlooked in previous assessments, as they were ASD conditions with less impact as the Asperger case.

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Appendix 5. PEDS Instrument: Questions on the concerns of parents about the development of their children. Translation of the PEDS Instrument (Parent's Evaluation of Development Status)[204]

QUESTIONS
– Please tell us if you are concerned about the way your child is learning, develops or behaves.
– Are you worried about how your child talks and makes sounds when talking?
– Are you worried about how your child understands what you say?
– Are you worried about how your child uses his or her hands and fingers to do things?
– Are you worried about how your child uses his or her legs and arms to do things?
– Are you worried about your child's behaviour?
– Are you worried about how your child interacts with other people?
– Are you worried about how your child is learning to do things for himself?
– Are you worried about how your child is acquiring skills in the nursery or at school?
– Please let us know any other concerns.

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Appendix 6. Instrument for assessing the risk of ASD: M-CHAT Assessment tool Format (for parents)

The information contained in this questionnaire is completely confidential.

Select, by **circling** the answer that you think best reflects how your child behaves NORMALLY. If the behaviour is not normal (for example, you have only seen it done once or twice) reply that the child does NOT behave this way. Please answer all questions.

1. Does he or she like to be swung, or that the adult does a "wheelie" by sitting him on the adult's back, etc.?	YES	NO
2. Does he or she show interest in other boys or girls?	YES	NO
3. Does he or she like standing on places such as chairs, stairs, and swings at the playground...?	YES	NO
4. Does he or she like playing "peek-a-boo" with an adult (cover his or her eyes and then uncover them, play at appearing and disappearing)?	YES	NO
5. Does he or she ever perform imaginative games, for example acting as if talking on the phone, feeding a doll, driving a car or something similar?	YES	NO
6. Does he or she usually point out with his or her finger to ask for something?	YES	NO
7. Does he or she usually point out with his or her finger to indicate that something catches his or her attention?	YES	NO
8. Can he or she play properly with small toys or pieces (i.e. cars, dolls, building blocks) without just sucking, shaking or throwing them?	YES	NO
9. Does he or she usually bring objects to show them to others?	YES	NO
10. Does he or she often look into your eyes for a few seconds?	YES	NO
11. Does he or she seem too sensitive to noises that are not very intense? (i.e. reacts covering his or her ears, etc.).	YES	NO
12. Does he or she smile when they see you or when you smile at him or her?	YES	NO
13. Can he or she copy or repeat gestures or actions that you do? (i.e. if you make a grimace he or she also does it)	YES	NO
14. Does he or she respond when called by name?	YES	NO
15. If you point with a finger at a toy across the room... does your child direct his or her eyes to that toy?	YES	NO
16. Has he or she learned to walk?	YES	NO
17. If you are looking at something closely, does your son or daughter stop to look at it too?	YES	NO
18. Does your son or daughter do odd movements with his or her fingers, for example, bringing them close to his or her eyes?	YES	NO
19. Does he or she try to make you pay attention to the activities he or she is doing?	YES	NO
20. Have you ever thought that your child might have a hearing loss?	YES	NO
21. Does your son or daughter understand what people say?	YES	NO
22. Does he or she sometimes stare into space or go from one place to another without purpose?	YES	NO
23. If your son or daughter has to face an unknown situation, does he or she look first to your face to know how to react?	YES	NO

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DATA TO BE COMPLETED WITH THE M-CHAT SCALE

Date

Name of person completing the questionnaire:

(Specify): Relationship to Child:

Mother Father Other

DETAILS OF THE CHILD

Name and Surname:

Date of Birth:

Sex: Male Female

Contact Number:

Address:

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Information for professionals

Cut-off points in the questionnaire to be considered a "failure" (in bold)
- Failure in at least three of the 23 elements (the yes / no answers in bold are considered a failure)
- Failure in at least two of the six critical elements (numbers 2, 7, 9, 13, 14, 15 in bold)

M-CHAT Valuation Instruments (Format for the professional)

The information contained in this questionnaire is completely confidential.

Select, by circling the answer that you think best reflects how your child behaves NORMALLY. If the behaviour is not normal (for example, you have only seen it done once or twice) reply that the child does NOT behave this way. Please answer all questions.

1. Does he or she like to be swung, or that the adult does a "wheelie" by sitting him on the adult's back, etc.?	YES	**NO**
2. **Does he or she show interest in other boys or girls?**	YES	**NO**
3. Does he or she like standing on places such as chairs, stairs, and swings at the playground?	YES	**NO**
4. Does he or she like playing "peek-a-boo" with an adult (cover his or her eyes and then uncover them, play at appearing and disappearing)?	YES	**NO**
5. Does he or she ever perform imaginative games, for example acting as if talking on the phone, feeding a doll, driving a car or something similar?	YES	**NO**
6. Does he or she usually point out with his or her finger to ask for something?	YES	**NO**
7. **Does he or she usually point out with his or her finger to indicate that something catches his or her attention?**	YES	**NO**
8. Can he or she play properly with small toys or pieces (i.e. cars, dolls, building blocks) without just sucking, shaking or throwing them?	YES	**NO**
9. **Does he or she usually bring objects to show them to others?**	YES	**NO**
10. Does he or she often look into your eyes for a few seconds?	YES	**NO**
11. Does he or she seem too sensitive to noises that are not very intense? (i.e. reacts covering his or her ears, etc.).	**YES**	NO
12. Does he or she smile when he or she sees you or when you smile at him?	YES	**NO**
13. **Can he or she copy or repeat gestures or actions that you do? (i.e. if you make a grimace he or she also does it)**	YES	**NO**
14. **Does he or she respond when called by name?**	YES	**NO**
15. **If you point with a finger at a toy across the room, does your child direct his or her eyes to that toy?**	YES	**NO**
16. Has he or she learned to walk?	YES	**NO**
17. If you are looking at something closely, does your son or daughter stop to look at it too?	YES	**NO**
18. Does your son or daughter do odd movements with his or her fingers, for example, bringing them close to his or her eyes?	**YES**	NO

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19. Does he or she try to make you pay attention to the activities he or she is doing?	YES	**NO**
20. Have you ever thought that your child might have a hearing loss?	**YES**	NO
21. Does your son or daughter understand what people say?	YES	**NO**
22. Does he or she sometimes stare into space or go from one place to another without purpose?	**YES**	NO
23. If your son or daughter has to face an unknown situation, does he or she look first to your face to know how to react?	YES	**NO**

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Appendix 7. Autonomous Scale for detection of Asperger syndrome and high-functioning autism

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Application Instructions

Each of the statements you will read below describes ways of being and behaving that may be indicative of Asperger syndrome or autism. These people usually have in one way or another features similar to those listed here, **especially after 6 years old**.

Please read each statement carefully, and consider to which extent you have observed the following behaviours in the individual whom you will report about by marking the appropriate response with the following criteria:

1. If the behaviour described in the statement does not correspond at all with the characteristics of the individual whom you are reporting about, mark the space provided for the category **"Never."**
2. If these features are observed sometimes, although not usually, mark the space provided for **"Sometimes."**
3. If the behaviour described is common, answer **"frequently."**
4. If he or she usually behaves as described in the statement, to the extent that anyone would expect him to behave in such a way, answer **"Always."**
5. It may happen that the descriptions refer to behaviours that occur in situations where you have never been present, for example: "Eats without any help," is a behaviour that can only be observed if you have had the opportunity to be present at lunchtime. If this is the case, mark **"Not observed"**.

Instructions for the correction

1. Rate the responses to each item observed as follows:
   - "Never": 1 point
   - "Sometimes": 2 points
   - "Often": 3 points
   - "Always": 4 points
2. Check if they meet the following two conditions:
   - The 18 items have been answered
   - No more than two items are marked with the answer "Not Observed"
3. If these two conditions are fulfilled, obtain the *direct text score* by adding the points of all the responses (the minimum score you can get in this sum will be 18 and the maximum 72).
4. If these two conditions are fulfilled, obtain the *average test score* by adding the points of all the responses and dividing them by the number of items answered (the minimum score you can

get is 1 and the maximum 4).

5. It is recommended to consult a specialist if the *direct score* obtained is 36 (or close to this value), and if the *average score* is 2 (or close to this value), see Note.

NOTE: Nothing prevents the professional from starting the questionnaire directly if the direct score does not reach 36 but is above 30 points (or if the average score is less than 2 but it is above 1.7); also, there are no psychometric reasons which prevent adopting a more demanding criterion (for example, taking a direct score of 40 as cut-off point, or an average score of 2.22). This scale is not a diagnostic tool in itself, but a tool to facilitate the detection of people who may have Asperger syndrome or autism not associated with intellectual disabilities and for whom it would be interesting to determine their clinical diagnosis and support needs as soon as possible.

Autonomous Scale

ITEMS	Never	Some times	Frequently	Always	Not observed
1. Has difficulty performing tasks where it is absolutely necessary to extract the main ideas of the content and ignore the irrelevant details (i.e. telling a story, describing a person...).					
2. Shows difficulty understanding the ultimate meaning of non-literal expressions such as jokes, idioms, requests made by asking questions, metaphors, etc.					
3. Prefers to do things alone rather than with others (i.e., playing alone or merely observes how others play, prefers to do homework or work tasks on his or her own).					
4. His way of initiating and maintaining interactions with others is strange.					
5. It is difficult to understand subtle facial expressions if these are not too exaggerated.					
6. Has trouble reading the proper sense of words or expressions whose meaning depends on the context in which they are used.					
7. Lacks initiative and creativity in the activities in which he or she participates.					

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8. Makes use of stereotyped or peculiar social formulas during a conversation (i.e., greets or says goodbye in a special or ritualised way, uses infrequent or improper courtesy formulas...)					
9. Finds it difficult to make friends.					
10. Conversation with him / her is laborious and hardly fluid (i.e., conversation topics are very limited, he or she takes too long to respond or does not respond to comments and questions put to him or her, says things that bear no relation to what has just been said....).					
11. Gives the impression of not sharing the same interests, tastes, hobbies, etc., with the group.					
12. Has difficulty to work effectively with others.					
13. His behaviour is naive (not aware of being deceived or teased, can not lie or hide information, can not disguise or conceal his or her intentions...)					
14. Makes idiosyncratic use of the words (i.e., uses uncommon words or with infrequent meanings, assigns very specific meanings to some words).					
15. Others have difficulty reading his or her emotional expressions and signs of empathy.					
16. Has trouble to understand fictional situations (films, stories, plays, stories, role plays...).					
17. Performs or attempts to impose complex routines or rituals that hinder daily activities.					
18. In games, sticks rigidly to the rules and is inflexible (i.e., does not support changes in the game, never cheats and is intolerant with others)					
AVERAGE SCORE:	DIRECT TOTAL SCORE:				

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DATA TO BE COMPLETED ON THE AUTONOMOUS SCALE

DATE OF APPLICATION:

PERSONAL DATA ON PERSON REPORTED

NAME:

DATE OF BIRTH: AGE:

SEX:

DATA OF REPORTING PERSON

NAME:

RELATIONSHIP WITH THE PERSON BEING REPORTED:

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Appendix 8

Parents' Information for a suspected social and communicative development problem



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Health Technologies Evaluation Unit.

Gran Vía, 27. 28013, Madrid. Spain

Produced by: www.cege.es - Eloy Gonzalo, 25, 1 º izda. 28010, Madrid

Parents' Information for a suspected social and communicative development problem

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What does it mean that there are suspicions that your child has a social and communicative problem?

You may have noticed some strange behaviour in your child. If he or she has older siblings, you have been able to compare that the acquisition of skills was not being the same as that of his or her brothers, nor does he or she do the same things as other children of his or her age. For example, he or she is more quiet or passive than other children, does not utter any words, just babbles or uses gestures to communicate, does not answer to its name, just smiles and expresses delight when attempting to share activities with him or her, seems indifferent to what surrounds him, etc.

You may have discussed it with other people (family, friends, and professionals) or you were simply waiting to see if it solved as the child was growing.

You may have not suspected the existence of any problem and, in a routine examination, his or her physician has informed you that something seems to be wrong in the development of your child.



Although not all children develop their skills at the same time, there are some age characteristic periods in which some of them are acquired. To serve as guidance for those periods, for example, the child should be able to hold his or her head up alone at 6 weeks old, smile at about two months, sit up without support before 9 months, etc.

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Your paediatrician can inform you about those moments in the development. Probably, from the child's birth, during routine visits, you will have been asked about the acquisition of these and other skills.

In case you have not suspected anything before and have discussed this issue in one of the visits to the Health Centre, you must rely on the conduct of professionals, and that in any case, the aim is to find out if there something different in the development of your child, what is happening, and what actions can be put in place quickly.

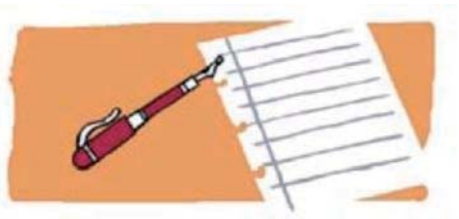
If your child attends a nursery or a school, it would also be useful to talk with the educators of the child to share with them your concerns. They can also provide a specific sign that could have been observed in the child's behaviour, which can guide the suspicions of the paediatrician.

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The fact that there is some warning sign does not necessarily mean your child has a development problem, however, it does indicate the need for further evaluation. Therefore, do not hesitate to discuss with your doctor any concerns, as he may indicate the completion of certain evidence, in order to identify possible causes that could explain the existence of these signals, among others, hearing problems, or delays in language development.

When these first warning signs are detected, it is possible that your doctor has asked you to answer a few questions about your child's behaviour, so as to confirm whether or not there is a high risk of having a problem and, if so, the child will be referred to specialised care for a fuller assessment and diagnose the presence or absence of a specific disorder.

Most importantly, what you must bear in mind is that the main objective is to rule out whether or not there is a problem, and have an accurate diagnosis as soon as possible to schedule the intervention that best suits the resources and characteristics of your child and the family.



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What is the procedure to follow?

If the possibility of having a problem of communication and social development exists, the child's paediatrician will send him to specialised care for further evaluation. The paediatrician will guide you on to what reference unit or specialty care centre you should address.

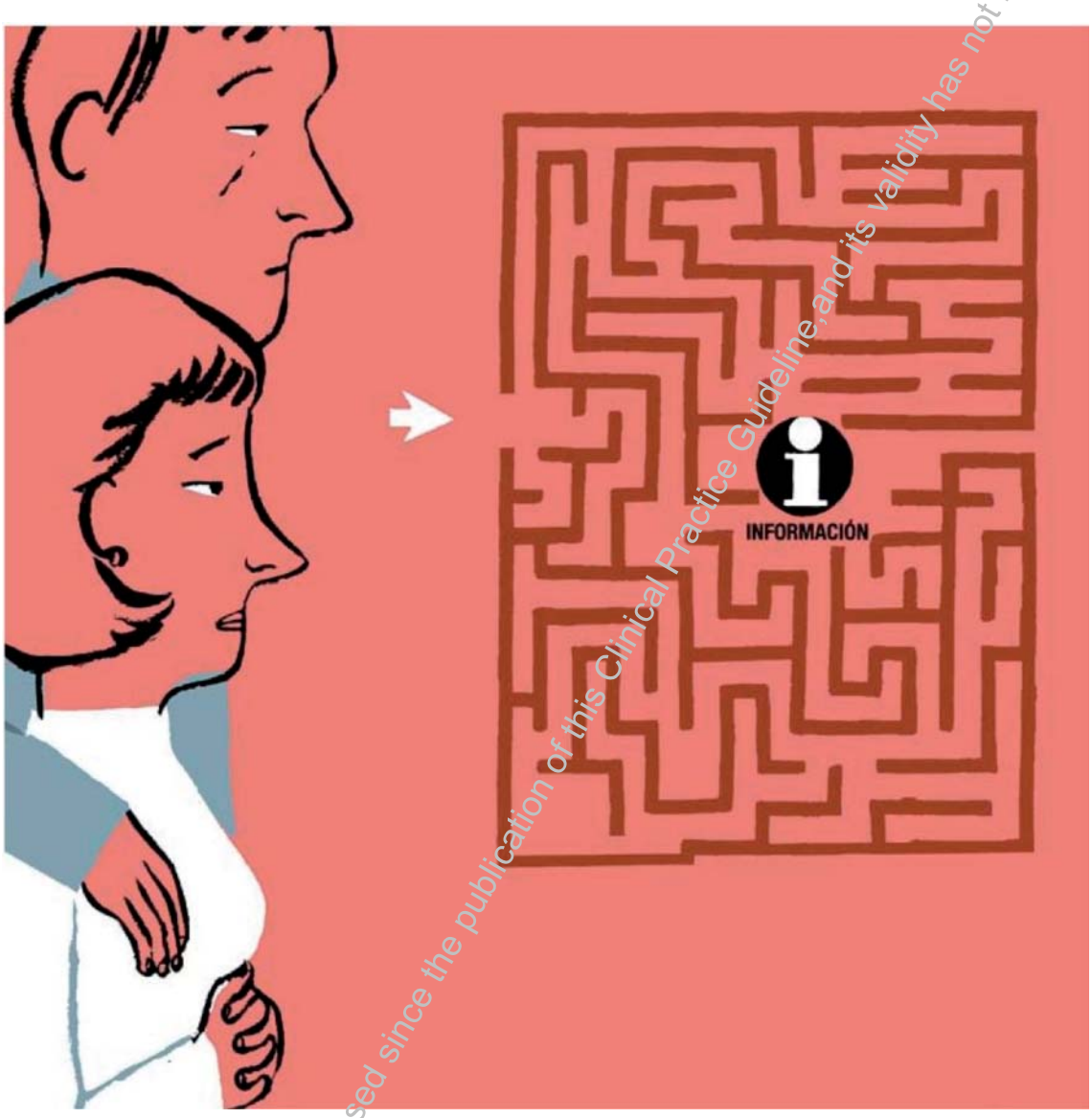
These centres have a multidisciplinary team that will assess several areas. As guidance, these teams can be composed of a paediatrician, a psychiatrist, a neurologist, a psychologist, a speech therapist, a nurse, an occupational therapist, a physiotherapist and a social worker.

This is a process based on observation of behaviour and psychological evaluation of your son in relation to his or her cognitive development, language and communication, and social skills. For this purpose, they may use more specific diagnostic procedures (mostly observational tests to assess your child's behaviour).

Specialists may also request some additional diagnostic tests such as blood tests, hearing tests and auditory evoked potentials that will rule out some metabolic diseases and hearing disorders.

It is also likely that the personal and family history of children are assessed, and that a genetic study is performed, since some of these disorders have identified a genetic basis, which can provide diagnostic information.

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This is a process that can last several weeks, and surely, the family will live with some level of stress. **In the moment they have an appointment with the reference centre or specialised care, the following questions may arise. We will try to answer these below:**

Approximately, how long will it take to get a diagnosis?

The total duration of the process is variable, and may be extended if there is need for additional evidence (audiometry, neuroimaging, genetic and other). In any case, once all the results are obtained, it will take no more than a few days to have the conclusions of the diagnosis. It is recommended that you are provided a written report explaining the results and guidelines to follow. Anyway, regardless of the time required to perform any additional tests, the clinical examination by professionals dedicated to this kind of problems will surely clarify your doubts.

What professionals will evaluate your child?

In general, the evaluations may be carried out jointly by several professionals who conduct interviews with families and children tests simultaneously. This is recommended for several reasons. First, because it allows to make the most of the time devoted to evaluation, since it takes information from two main sources (family and children) at the same time.

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Second, it increases safety by allowing clinical trial to compare the observation of several different professionals. Do not forget that the diagnosis is made based on the observed behaviour of the child, taking into account the history of development that the family facilitates. Sometimes, it is difficult to interpret the information, for this reason it is recommended to perform some contrast views between the clinical opinions of various professionals.

Normally, the professionals who will conduct the interviews and tests to the child belong to the health care field (clinical psychologists, paediatricians, psychiatrists, neurologists, and nurses), the education field (guidance teams and educational assessment) and the social field (assessment and counselling centres.)

What kind of evaluation will be performed?

The tests made to the child will focus on the observation of your child's behaviour (how he or she mixes and communicates with adults and peers), the way in which he or she gets things he or she wants (if otherwise requested or taken by himself), how he or she gets involved in games, if he or she shows some behaviours that may seem unusual (uses objects or parts thereof in a repetitive manner, insists on putting the toys in the same position, does not change in their distribution...).

It is also likely to carry out some tests to help to identify the child's cognitive skills (attention, understanding, abstraction, memory, ability to learn, communicate, and others) and the level of development.

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It will be particularly important to explore the child's competences related to language and communication skills as well as other aspects of the development, as well as other adaptive skills that enable the successful adaptation to the environment, depending on what is expected for the age of the child.

What kind of evaluation will be performed?

Fundamentally, information on how the overall development of your child has been up to now and what is his or her behaviour in everyday life and everyday situations, as it is possible that during the evaluation he or she does not reveal such behaviour for being within an unfamiliar environment. A wide-ranging interview is performed to parents about child development, and on the possible signs that have appeared, which may help to confirm or refute the diagnosis. The problems of development may go unnoticed at very early ages, so you have to review those indicators or warning signs that might appear or are seen even before 12 months of age. The cooperation between the professional conducting the interview and the parents will help to identify whether these signs have been present or not, this being essential information for the diagnosis.

In this history of the development, possible family history with characteristics similar to those presented by the boy or girl, or some other that can be associated will be consulted. Issues related to pregnancy, childbirth and the early stages of the child's development, including the results of the tests performed to newborns (Apgar score, phenylketonuria, thyroid function...) will also be assessed.

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You will also be consulted on major development milestones, both in relation to motor aspects (at what age he or she began to sit, when he or she started to walk...), as well as social interaction and communication (what is the relationship with other children or adults, if he or she has started to talk and how is the language used, if there is any kind of loss of language skills or other skills acquired...).

In relation to these aspects, the elements that constitute the major warning signs that guide the specific evaluation of the developmental disorders will be explored in detail. Amongst others, the information on the development of communication skills and interpersonal relationships of children will be particularly important, paying special attention to whether the child communicates / interacts / plays / competes / shares or not with others, and how he or she does it. For example, if there is delay in language or in the case where he or she does not talk, if he or she uses other means to communicate, how the child expresses what he or she wants, whether he or she answers or not when asked, if the child acts in an inconsistent manner regarding the feelings of others such as anger or joy, if he or she starts social interactions and shares them with others or if the child undertakes them only when he or she needs something, if he or she repeats words out of context, if he or she keeps the people's looks and follows them.

You will also be asked whether the child has some interests or specific concerns to which he or she devotes much time which could interfere with the performance of other activities and which are unusual, by the intensity with which they are presented, for their content or the subject. The child's abilities to accept changes in his or her routines, and if there is any insistence on always performing the same activities in a repetitive manner with difficulty to accept changes in them (for example, change the order of the toys, or the journey to school...) will also be assessed.

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Finally, consideration will be given to the way in which the child plays, what kind of games and toys are used and if used in a particular or unusual way. Also, taking into account the age, if he or she pretends to play imaginative or fiction games, that is, plays with something as if it were something else knowing it is not, for example, "giving food to dolls", "act as if a block of wood was a car", "talking on the phone using a spoon," "mimics riding a horse with a broom stick"...

What should you prepare for the appointment?

In the event that you have already gone to visit other professionals, it is advisable to provide the team who is going to perform the evaluation reports with previous results, if possible, before going to the appointment with them. This will allow better a planning of the evaluation process, avoiding duplication of evidence, and dedications to others that may be relevant, but which have not been carried out yet. It is also interesting to provide the reports of the nursery or school, as here it is very likely to detect signs of the problem, if it exists.

It is also important to prevent what may be the needs of your child during the evaluation. Bring with you objects or toys that can entertain him or her during the breaks, or which can be used once the test is finished.

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Bear in mind that your son or daughter goes to an unknown place, with people who he or she has never seen. It is recommended that, as far as possible, you inform the child of where he or she is going, the people he or she is going to meet and what is going to happen there, because, as you can see, he or she does not usually tolerate changes in routine. To do this, you can use photographs of the place where the assessment is going to be conducted, or of the persons who are going to be with him or her. If it were possible, it would be advisable to visit the place before conducting the tests, so that it is not be completely unknown to the child.

Will I be with my child during the evaluations?

It is very unlikely that any member of the family can stay with the child throughout the evaluation. It is also possible that the people who perform the tests request your collaboration in some moments, especially for tasks relating to social aspects and games.

In any case, consult the doubts you may have about the procedures and if you can participate in the evaluation with the team of professionals while they are performing it. It is important to know which aspects are being explored and how your son or daughter responds during the evaluation. This will make it easier for the child to feel familiar with the concepts and elements that are described in the diagnostic report, where the findings will be presented in detail.

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How much time is needed in the evaluation process (observation of behaviour)?

The assessment tests vary in length, and this depends on several factors, especially on the fatigue and the collaboration of the child. It is necessary to devote some time for the child to become familiar with the environment and the people who will perform the tests, and it is possible that these have to be carried out in several sessions, which will probably be developed in different days.

In other cases, the assessment may last more than a whole morning, afternoon or even a day, providing the necessary breaks, being carried out continuously until the necessary information is obtained.

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Dealing with health professionals

Since your child will be referred to specialised care, he may have to go through various consultations of professionals from different fields and health, educational and social specialties, and in many cases, you will serve as a unifying and communication bridge between them. These moments can be very stressful for parents, especially when they already suspected that something was different in the development of their child and they want to have a clarification on the matter.

During this evaluation process, your child will be treated by different professionals, so you may find useful the following **suggestions to facilitate relationships and communication with the professionals who will care for your child:**

- It is good *to show emotions and concerns to the professionals*. This helps professionals to improve understanding of your situation.
- *Communicate your stress level and share the complex situations* which your child is going through.
- Despite the experience of other professionals, *you are the ones who know your child best and have valuable information that should be shared*. Avoid preconceived ideas derived from feedback received previously by the family or even other professionals. *Express openly your fears and doubts.*

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- As the information professionals provide you with can be difficult to assimilate and to remember in the early stages, you should *not come alone to the interviews*. Try to be accompanied by a partner, relative or friend who could help you understand the information better.

- *Make a list of questions* which you may have to take advantage of the visits to the physician and try to deal with them with the professionals.

- Request that the information be provided in a simple language, avoiding jargon. Remember that at the end of the process you will be provided with a report with all the tests, the results obtained the final diagnosis, proposed treatment guidelines, reviews, etc... You must keep this file.

- If you do not agree with the information received, you *may request a second opinion*.

- *Maintain communication and clarification of emerging issues* with professionals.

- Try to write things, whether the information received or scheduled appointments, names, dates, even phone calls. *Maintain, a detailed agenda, wherever possible.*

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www.madrid.org / lainentralgo

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Appendix 9

Parents' Information for a diagnosed Autism Spectrum Disorder (ASD)



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This informative document for parents is part of a Clinical Practice Guideline (CPG) developed under the Quality Plan for the National Health System of the Ministry of Health and Social Policy. 2009.

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Health Technologies Evaluation Unit.

Gran Vía, 27. 28013, Madrid. Spain

Produced by: www.cege.es - Eloy Gonzalo, 25, 1 º izda. 28010, Madrid

Parents' Information for a diagnosed Autism Spectrum Disorder (ASD)

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Introduction

Despite the logical sense of relief that takes place when you are given an explanation of what happens to your child and to know, finally, that your concerns were founded and were not product of your imagination, acceptance of the diagnosis will require time and will convey to a process full of ups and downs. It is completely normal that the confrontation with the reality of a child who will have development and different needs than expected compared with other children of their age, will cause a conflict with the expectations set upon him and throw a great concern about his or her future.

During this process different emotions may appear, from the initial denial, through feelings of anger, resentment and depression, until the moment of acceptance. According to the families who already have a child with this disorder, the first moments after receiving the diagnosis are the most difficult, but with time and adequate support, most parents feel better. The acceptance of their child as he or she is, with his or her own characteristics, strengths and weaknesses, like any other child, will help to provide the support to develop their skills and overcome any difficulties you may have. It will also help the rest of the family and friends to learn to know the child and accept him or her as he or she is. The sense of control over the situation improves safety and the coping ability of parents, therefore, information on diagnosis and the way in which it can influence your child's life is extremely useful.

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After obtaining a diagnosis, your doctor will ask you for that information, so that he can set up visits and the relevant follow-up for your child. Moreover, he may also take into account the needs of surveillance and the specific routine health care of your child or the needs associated to the diagnosis that may occur in the future.

If you are concerned that in the case of having more children, some of them may also suffer from an ASD, do not hesitate to request information. ASDs have a genetic basis and sometimes are associated with some genetic syndromes as well, thus that information can help you make a decision if you want to have more children. It is important to know that if you have a child with ASD, the odds of having other children with this type of diagnosis or other developmental disorders are higher compared to the general population.

Overview of Autism Spectrum Disorders (ASD)

ASDs are a group of disorders that affect child development. These changes occur because there is a chronic neurological disorder base. The way in which these changes are manifested varies greatly from child to child, hence the fact that these are called a "spectrum" or "continuum of disorders," i.e., that there are different ways in which the symptoms of this type disturbances occur, and their severity varies from case to case. Therefore, the term covers from more severe cases to mild cases with very good prognosis. In addition, ASDs are more common in boys than in girls and occur equally in any cultural and social group.

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Disorders will be presented in three main areas of development: changes in their communication and language skills, impaired social interaction skills, as well as the presence of a very restricted repertoire of interests and behaviours, which limit the variety of activities and daily behaviours of the individual. Sometimes the child may also present some unusual features, like a great sensitivity to certain auditory stimuli (sounds), tactile (fabrics, textures and even to smell...), and even to different flavours.

In most cases, children show a normal physical appearance although they may present different changes in their behaviour. Therefore, until certain characteristic moments of development are reached, and differences in relation to the ordinary development are observed, no warning signs that may indicate the presence of an ASD are perceived.

Sometimes, ASDs may be accompanied by other associated health problems such as epilepsy, cognitive impairment*, movement problems, eye and hearing disorders, hyperactivity, anxiety, insomnia and others. In addition, as from adolescence, they can begin to show other associated mental health disorders such as obsessive compulsive disorder, anxiety, depression and other disorders in their mood, especially in children with autism without associated intellectual disabilities.

* Cognitive impairment can affect skills such as attention, comprehension, memory, learning ability, communication and decision making.

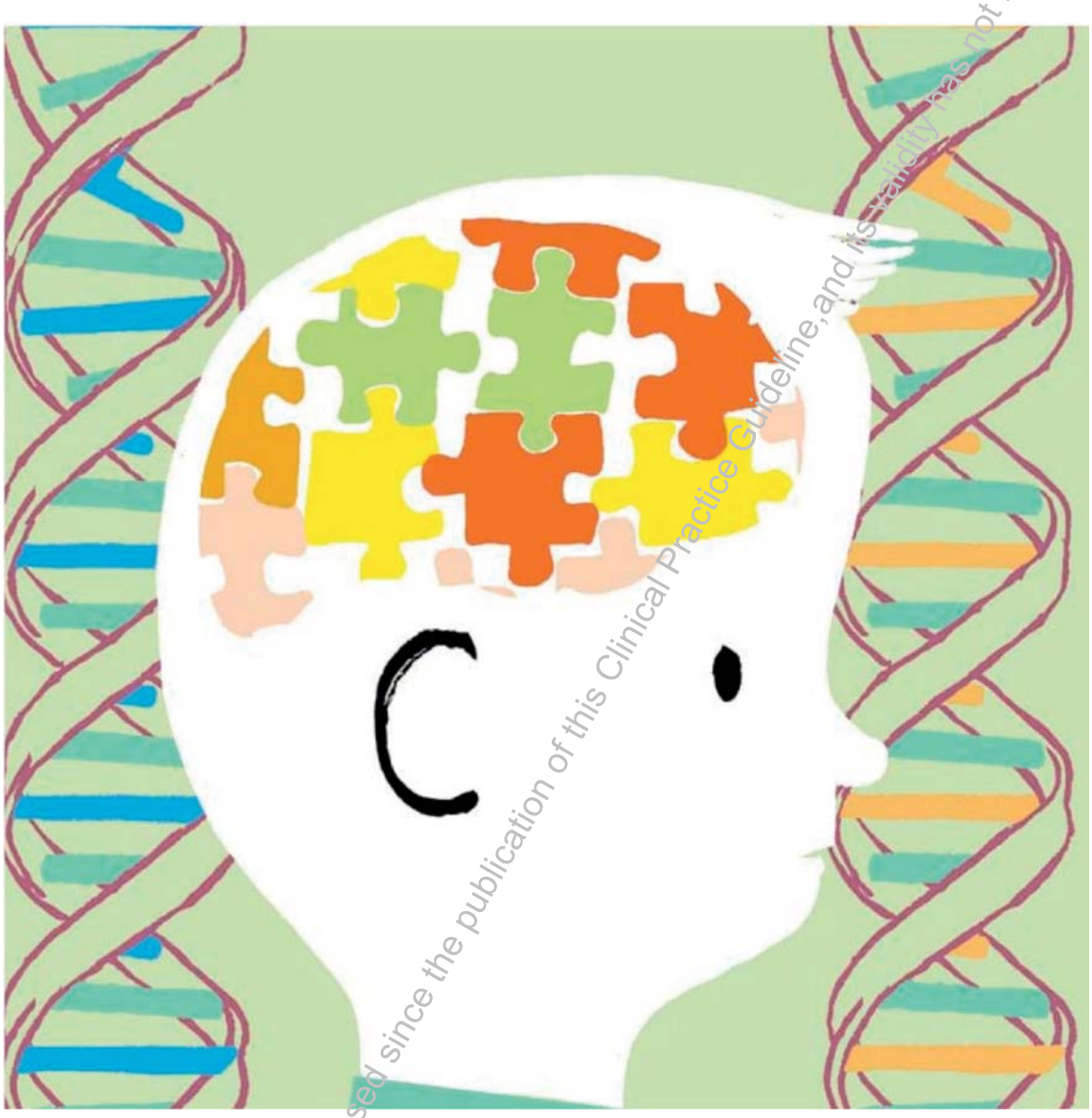
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ASDs are chronic conditions, which cannot be cured, i.e., they do not disappear during one's life. However, by providing intervention and appropriate support, children can develop their own skills in different areas (social, communication, and other daily routines.) There is also a decreasing trend of behavioural difficulties with the course of aging.

Bear in mind that children with ASD have the same needs of love and family support than other children, but sometimes have difficulty expressing it. Each child has a unique personality and expression of the disorder, therefore, is different in each case. The sooner we know the strengths and weaknesses of the child, as well as the understanding of their characteristics and needs, specific actions to encourage their development and social integration may also be fostered.

It is essential to know which treatment and intervention approaches that have been scientifically proven and its effectiveness has been tested, are tailored to the needs of your son or daughter, so we advise you to trust the experts when it comes to receiving advice. Nowadays, individualised education and specialised support are the most effective tools to encourage the development of your son or daughter. However, quite often, some therapeutic approaches are offered which defend magic, miraculous and scientifically unproven cures; these are ineffective and will only create more frustration to parents and an unnecessary financial expenditure.

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What is the origin of ASDs?

There is a genetic component that may act on the environmental factors that develop these disorders. Some parents, especially mothers, tend to think that something must have occurred during pregnancy, childbirth etc. which has caused the disorder, which is uncertain. This idea increases even more the level of distress experienced by parents and their feelings of guilt about what happened. In any case, it must be clear that what happens to children with an ASD is not the responsibility of their parents, there was no way to prevent it and, of course, has nothing to do with their upbringing.

Social stereotypes on ASDs

Despite how much progress has been made in recent years in the knowledge about ASD, there are still many socially widespread misconceptions about people with these disorders. Autism is a disorder which is particularly difficult to accept from the perspective of parents, because children do not show any physical characteristics that indicate a change in their development, and for the rest of the people, apparently, there is no explanation for their behaviour.

The most widespread misconceptions about ASD are discussed below. Each idea is accompanied by an explanation of why they are wrong:

- **Stereotype 1: They behave in this way because their parents do not love them or have spoilt them.** Autism is a chronic neurological disorder that has nothing to do with the upbringing received. Children can not avoid the behaviours they show, but many of them may be modified through appropriate interventions

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- **Stereotype 2: All children with ASD have mental retardation.** Although there is often cognitive impairment, some children with ASD have normal IQ scores and some even above average, and others have special skills in areas such as mathematics or numeracy, music, painting, etc., though the majority do not have these features.

- **Stereotype 3: They can not communicate, do not look in the eyes, do not smile or respond to signs of affection.** The social skills of children with ASD are affected in a variable and different way in each case. Some children are diagnosed late in adolescence and beyond, since the existing alterations are difficult to detect because they can present an apparently appropriate language and behaviour. In addition, children with ASD have difficulty interpreting and recognizing emotions in others and sometimes emotional expressions do not fit the situations that occur and are different from what is socially expected. In any case, they are sensitive to signs of affection, and express their feelings and their preferences, but sometimes do so in a manner different from that of other children of their age.

- **Stereotype 4: The origin of the problem is related to the administration of vaccines, food intolerance (gluten, casein), or heavy metal poisoning (mercury, lead).** These theories currently lack scientific support and have been refuted.

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Remember that your child is not an "autistic child" but a "child with an autism spectrum disorder" that is, your child has a unique personality, just like any other child, which is why the expression of this disorder in your son or daughter, the symptoms and their present or future capabilities will be different to other children with ASD.

What are the external and internal conditioning factors that negatively or positively affect the prognosis?

The progress or prognosis of children with ASD is highly variable and will depend on a number of factors in each case, as is the level of cognitive functioning, the absence or presence of associated disorders (organic brain disorder, epilepsy, sensory abnormalities...), the level of language development and social and communication skills as well as other external factors such as the family, educational and social support provided. These environments will influence positively or negatively on the development of the child's abilities and, therefore, in his or her longer term prognosis.

Despite the chronicity of ASDs, appropriate educational interventions and appropriate support (individual and family) are conducive to the development of children's abilities; they can achieve significant learning which is necessary for their personal development and quality of life. Parents, and by extension the rest of the family, must be an active and integral part of the interventions and contribute to the learning of children.



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The process which they have to go through with their child is a hard one, sometimes with many troubles, but also full of satisfaction for the achievements of his son or daughter. There will be people like you at your side, just like you, they are at various stages of this process and can guide you. Do not hesitate to seek the support of both, professionals and your family, friends, other families of children with ASD or associations whose experiences will enrich you.

Dealing with health professionals

During the diagnostic process and after receiving the diagnosis that your child suffers from an ASD, you will have to deal with a large number of professionals from different fields and of health, educational and social specialties, and in many cases you will serve as union and communication bridge between them. The follow-up visits as well as the regular assessments and coordination with all the professionals who will care for your child can be stressful for parents, especially for those who play the role of primary caregiver.

With time, you will become experts in the field and all this will be easier, but, especially in the early stages, the use of these **suggestions to facilitate the relationship and communication with professionals who care for your child can be useful:**

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- It is good *to show your emotions and concerns to the professionals*. It helps professionals to improve understanding of your situation.

- *Communicate your stress level and share the complicated situations* which your child is going through.

- Despite the experience of other professionals, *it is you who know your child best and have valuable information which ought to be shared*. Avoid preconceived ideas derived from feedback received previously by the family or even other professionals. *Express openly your fears and doubts.*

- As the information which professionals provide you with can be difficult to assimilate and to remember in the early stages, you should *not come alone to the interviews*. Try to be accompanied by a partner, relative or friend who could help you understand the information better.

- *Make a list of questions* which you may have to take advantage of the visits to the physician and try to deal with them with the professionals.

- *Request that the information be provided in a simple language*, avoiding jargon. Remember that at the end of the process you will be provided with a report with all the tests, the results obtained the trial diagnosis, proposed treatment guidelines, reviews, etc. You must keep this file in case it is necessary afterwards.

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- If you do not agree with the information received, you *may request a second opinion.*
- *Maintain communication and clarification of emerging issues* with professionals.
- Try to write things, whether the information received or scheduled appointments, names, dates, even phone calls. *Maintain a detailed agenda, wherever possible.*
- If your child needs health care, *seek information about his or her needs and peculiarities before the visit* so that professionals can prepare this consultation.
- Do not forget to get in contact *with associations of parents* who can guide you thanks to their experience and about the professionals who work with them.

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Resources

Associations of families and individuals with ASD

FedFederación Española de Padres/Tutores de Personas con Autismo (FESPAU)
C/ Navaleno, nº 9, 28033 Madrid
Telephone: 91 766 00 18 / 91 766 00 18 / Fax: 91 767 00 38
Email: autistas@fespau.es
http://www.fespau.es/webfespau.html

Confederación Autismo España (CAE)
C/ Eloy Gonzalo, nº 34, 1º, 28010 Madrid
Telephone: 91 591 34 09 / Fax: 91 594 18 31
Email: confederacion@autismo.org.es
http://www.autismo.org.es

Federación Asperger España
C/ Foncalada, nº 11, esc. izq. 8º B
33002 Oviedo - Asturias
Telephone: 639 363 000 / Fax: 984 16 12 77
Email: info@asperger.es
http://www.federacionasperger.es

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Documents of interest

A child with autism in the family
Basic Guide for families who have received a diagnosis of autism for their child. Ministry of Labour and Social Affairs. Universidad de Salamanca 2007. Available at:
http://sid.usal.es/idocs/F8/FDO20581/familia_autismo.pdf

Manual for the first 100 days
A handbook to assist families to get the critical information they need during the first 100 days after a diagnosis of autism. (Spanish version). Autism Speaks 2008. Available at:
http://www.autismspeaks.org/docs/family_services_docs/manual_ de_ los_100_dias.pdf

Information available in Websites in Spanish on ASD

http://www.gat-atenciontemprana.org/1_AtencionTemprana/index.htm
Federación Estatal de Asociaciones de Profesionales de Atención Temprana-GAT (National Federation of Early Care Professionals Associations –GAT)

http://www.feaps.org
Confederación Española de Organizaciones en favor de las Personas con Discapacidad Intelectual- FEAPS (Spanish Federation of Organisations for People with Intellectual Disabilities-FEAPS)

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http://www.rpd.es
Real Patronato de Discapacidad
(Royal Board on Disability)
C / Serrano, 140, 28006 Madrid
Phone: 91 745 24 49 / Fax: 91 411 55 02
Email: rp@futurnet.es

http://sid.usal.es
Servicio de Información sobre Discapacidad
(Information Service on Disabilities)
University of Salamanca. Ministry of Health and Social Policy

http://www.isciii.es/htdocs/centros/enfermedadesraras/autismo/objetivos.jsp
Instituto de Salud Carlos III. Group of Experts on ASDs. Large number of publications, documents and resources

http://www.cdc.gov/ncbddd/Spanish/spautism.htm
The National Centre for Birth Defects and Developmental Disabilities (NCBDDD) is part of the Centre for Disease Control and Prevention (CDC) and has useful information on autism:
1600 Clifton Rd, Atlanta, GA 30333, USA
Enquiries: (404) 639-3534 / (800) 311-3435

http://espanol.ninds.nih.gov/trastornos/autismo.htm
The National Institute of Neurological Disorders, United States. It has useful information on ASDs in the unit on Neurological Disorders.

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http://autismo.uv.es
The Group of Autism and Learning Disabilities. Website of the University of Valencia. They have very useful technological tools adapted to the needs of people with autism, their professionals and family, applications on mobile devices, accessibility to Web sites and others

www.fundacionadapta.org
Fundación Adapta. Collaborates with the previous link. It is an organisation that promotes the development of new technologies for education, communication and self-determination of people with autism and learning difficulties. For example, the media resource "Miradas de Apoyo". More information at: https: / / www.miradasdeapoyo.org

Information available on international websites on ASD

http://www.nas.org.uk
National Autistic Society
The National Autistic Society 393 City Road, London, EC1V 1NG, United Kingdom
Phone: +44 (0) 20 7833 2299 / Fax: +44 (0) 20 7833 9666
Email: nas@nas.org.uk
Autism Helpline 0845 070 4004

http://www.worldautismorganization.org/es/introduccion.html
World Autism Organization WAO
Avenue E. Van Becelaere 26B (bte. 21) 1170 Belgium

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http://www.autismeurope.org
Autism Europe. 39 Rue Montoyer
1000 Bruxelles, Belgium
Phone: +32 (0) 2 675 75 05 / Fax: +32 (0) 2675 7270

http://www.autismspeaks.org
Autism Speaks / National Alliance for Autism Research
2 Park Avenue 11th Floor
New York, NY 10016
Phone: 212-252-8584 / California: 310-230-3568 Fax: 212-252-8676
Email Address: contactus@autismspeaks.org

http://www.nichcy.org
National Dissemination Centre for Children with Disabilities
U.S. Dept. of Education, Office of Special Education Programs
PO Box 1492 Washington, DC 20013-1492
Phone: 800-695-0285 / Fax: 202-884-8441
E-mail: nichcy@aed.org

http://ani.autistics.org
Autism Network International (ANI)
PO Box 35 448
Syracuse, NY 13235-5448
Email: jisincla@mailbox.syr.edu

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Multimedia Resources

Short film Mi hermanito de la luna (Little brother of the moon)

This beautiful short film, almost 6 minutes long, was created by Frédéric Philibert, parent of a child with autism. It shows the impressions of a girl, his eldest daughter, about his autistic brother. Released in France by the Orange Foundation; the Orange Foundation in Spain has made a subtitled version in Spanish. Link: http://www.fundacionorange.es/fundacionorange/comunicados/2008/petit_frere.html

Project "Pictogram Room" by the Orange Foundation

Pictogram Room (room for pictograms) of an augmented reality room to teach people with ASD to understand pictograms. This project is developed by the Orange Foundation in collaboration with the Group for Autism and Learning Disabilities at the Robotics Institute of the University of Valencia and the University of Weimar Germany in order to create software that enables people with autism to understand the meaning of pictograms. In the experimental evaluation of the developments two users' associations are also cooperating, the Asociación Autismo Ávila and the Asociación Autismo Burgos. The project, which started at the end of 2007, is planned to last three years. More information on: http://www.fundacionorange.es/fundacionorange/proyectos/proyecto_pictogram.html

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"Azahar" Project by the Orange Foundation

The Azahar Project, developed by the Group for Autism and Learning Disabilities at the Robotics Institute of the University of Valencia, aims to develop a combination of communication, entertainment and planning applications, executed through the mobile phone, to help improve the quality of life and independence of people with autism. It is one of the first approaches to the use of applications based on mobile telephony developed specifically for people with autism or other pervasive developmental disorders. The use of the mobile phone as a tool of communication and planning was considered optimal because, today, it is an object of widespread use. More information on: http://www.fundacionorange.es/fundacionorange_proyectos/proyecto_azahar.html

ZAC Browser - Zone for Autistic Children

Free web browser that allows your child to interact directly with games (many games and interests) and activities (focused on many interests) that cater specifically to children who exhibit characteristics of autism spectrum disorders. ZAC is the first web browser specifically developed for children with autism and also provides an excellent forum for parents, carers, teachers and others to share their experiences, tools and resources, and to unite as a caring, compassionate, and extremely knowledgeable community. Link and download on: http://www.zacbrowser.com/es

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http://miguel-gallardo.blogspot.com/2007/12/maria-y-yo.html

Blog by Miguel Gallardo, author of the comic *Maria and I*, in which the author recreates everyday situations with his daughter Maria, sharing with the reader the subject of autism in a natural, delicate and especially humorous way.

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Easy reading and adapted tales for children with autism

The Publisher Kalandraka, in partnership with the Asociación de Tratamiento de Autismo BATA (Association on the Treatment of Autism), has launched a collection of stories specifically aimed at children with special educational needs under the name of Malakin:

The Ugly Duckling
Author: Ana Sande (Illustrator) - Asociación de Tratamiento de Autismo BATA

The White Rabbit
Author: Ballesteros, Xosé and Bayonne - Asociación de Tratamiento de Autismo BATA

The Vain little Mouse
Author: Lopez Parreno, JA and Bayonne - Asociación de Tratamiento de Autismo BATA

Chivos chivotes
Author: Federico Fernández

More about Publisher Kalandraka on the website:
www.kalandraka.com
Phone: +34 986 752 485 / +34 986 860 276 ext. 2
Email: comercial@kalandraka.com

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Books

Comprender el autismo: un recorrido por los trastornos del espectro autista y el síndrome de Asperger a lo largo de todas la etapas escolares hasta la edad adulta

(Understanding autism: a journey through autism spectrum disorders and Asperger syndrome during all stages of school to adulthood)

Author: Stanley Greenspan
Editor: RBA libros, S.A. Date of issue: 2008

María y yo
(Mary and I)
Authors: María Gallardo y Miguel Gallardo
Editor: Astiberri. Date of issue: 2008

¿Qué le pasa a este niño? Una guía para conocer a los niños con discapacidad
(What's wrong with this child? A guide for children with disabilities)
Authors: Ángels Ponce y Miguel Gallardo
Editor: Serres. Date of issue: 2005

Mi familia es diferente: cuaderno de actividades para hermanos y hermanas de niños con autismo o síndrome de Asperger
(My family is different: activity book for brothers and sisters of children with autism or Asperger syndrome)
Author: Brock, C.
Translation: Javier Arnáiz y Javier Guadilla
Publisher: Burgos: Autismo Burgos, 2008 Notas: Editada en colaboración con la National Autistic Society (NAS)

Los Hermanos de Niños con Autismo
(The Siblings of Children with Autism)
Autor: Harris, Sandra L.
Publisher: NARCEA, S.A. DE EDICIONES, 2000. Date of issue: 2000

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Mi hermano tiene autismo
(My brother has autism)
Authors: Gónzalez Navarro, Ana y Labat Gronchi, Victoria
Publisher: Edited by the Minisrty of Work and Social Affairs (Publishing department).
Date of issue: 2008

El niño pequeño con autismo
(The kid with autism)
Authors: Riviere, A. y Martos, J. (Comp.) Publisher: Autor-Editor. Date of issue: 2000

Imagínate... que es un diario
(Imagine…. it's a diary)
Authors: De Susan M. Noonan y María Eugenia Leitao
Publisher: SAC-O - Stimulating Activities Company, SL. Date of issue: 2004

Los niños pequeños con autismo. Soluciones prácticas para problemas cotidianos
(Young children with autism. Practical solutions for everyday problems)
Authoes: Juan Martos Pérez
Publisher: CEPE. Date of issue: 2008

Hablando nos entendemos los dos
(We understand each other by talking)
Author: Manolson, Ayala
Publisher: Publicación del Centro Hanen. Date of issue: 1995

Déjame que te hable de los niños y niñas con autismo de tu escuela
(Let me tell you about children with autism from your school)
Authors: Juana Mª Hernández Rodríguez, Arantxa Martín Pérez y Beatriz Ruiz Casas
Publisher: Teleno ediciones. Date of issue: 2007

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www.msps.es
www.madrid.org / lainentralgo

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Appendix 10. Glossary and Abbreviations

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Glossary

AGREE (Appraisal of Guidelines, Research and Evaluation for Europe): International Initiative to facilitate the design and evaluation of clinical practice guidelines.

AVAD (DALY: disability adjusted life year) measure of the global burden of disease that reflects the number of years a person could have lived, years lost due to premature mortality and the years of productive life lost due to disability.

Case-control Study: This study identifies people with a disease (cases), for example lung cancer, and compares them with a group without the disease (control). The relationship between one or more factors (i.e., tobacco) associated with the disease is examined by comparing the frequency of exposure to these or other factors between cases and controls.

Case Series: Analysis of series of patients with the disease.

Cochane Library: Database of effects produced by the Cochrane Collaboration, composed among others by the original systematic reviews of this organisation.

Cohort study: It consists in following one or more cohorts of individuals having different degrees of exposure to a risk factor in those people where the onset of the disease or condition being studied is measured.

Comorbidity: The presence of one or more disorders (or diseases) in addition to the primary disease or disorder.

Cross-Study Description: It is the one that describes the frequency of an event or exhibition at any given time (single measurement). It examines the relationship between a risk factor (or exposure) and effect (or outcome) in a defined population and at a given time (a cut-off). Also known as prevalence studies.

DSM-IV: Fourth edition of the Diagnostic and Statistical Manual of Mental Disorders by the American Psychiatric Association. This is a classification of mental disorders in different types based on sets of criteria with defining features. It is created for clinical use, education and research. Its purpose is to provide clear descriptions of diagnostic categories, so that clinicians and researchers can diagnose, monitor and exchange information and deal with various mental disorders.

Medline: Database predominantly clinical produced by the National Library of Medicine, USA available on CD-Rom and the Internet (PubMed).

Meta-analysis: A statistical technique that allows integrating the results of different studies (diagnostic tests, clinical trials, cohort studies, etc.) into a single estimate, giving more weight to the results of larger studies.

Morbidity: disease or frequency of a disease occurring in a population.

Mortality: Death rate or the number of deaths for a particular disease in a group of people and a certain period.

NICE (National Institute for Clinical Excellence) is part of the NHS (National Health Service). Its role is to provide doctors, patients and the general public the best available evidence, primarily in the form of clinical guidelines.

Phenotype: It is the expression of the genotype in a given environment. Phenotypic traits include

physical and behavioural traits. It is important to highlight that the phenotype can not be defined as the "visible manifestation" of the genotype, because sometimes the characteristics being studied are not visible in the individual, such as the presence of an enzyme.

Pleiotropy: Are the multiple phenotypic expressions that occur as a result of the expression of a gene, i.e., that a gene can have more than one function.

Prevalence: The proportion of people with a finding or disease in a given population at a given time.

Qualitative research: A methodology that includes a plurality of theoretical trends, methods and techniques, and is characterised by a study on the phenomena in their natural context, trying to find the meaning or interpretation of them from the meanings people attach to them. To do this it uses empirical materials (interviews, observations, texts, etc.) that can describe best the routine and problematic situations and what they mean in the lives of individuals.

SIGN (Scottish Intercollegiate Guidelines Network): Scottish multidisciplinary Agency that produces clinical practice guidelines based on evidence as well as on methodological papers.

Systematic review (SR): A review, in which evidence on a subject has been systematically identified, evaluated and summarised according to predetermined criteria. You can include the meta-analysis or not.

Abbreviations

AEPAP: Spanish Association of Primary Care Paediatrics

AEPNYA: Spanish Association of Child and Adolescent Psychiatry

AETAPI: Spanish Society of Professionals in Autism

AGREE: Appraisal of Guidelines Research and Evaluation

ANESM: National Association of Mental Health Nursing

ASD: Autism Spectrum Disorders

CAE: Spanish Autism Confederation

CAST: Childhood Asperger Syndrome Test

CGH-arrays: Microarrays of comparative genomic hybridisation of the complete genome (diagnostic test)

Chat: Checklist for Autism in Toddlers

CINAHL: Cumulative Index to Nursing & Allied Health Literature

CINDOC: Centre for Scientific Information and Documentation from the Spanish National Research Council

CPG: Clinical Practice Guideline

DALY: Disability Adjusted Life Year

DARE: Database of Abstracts Reviews Effects

DC 0-3R: Diagnosing Infant and Early Childhood Mental Health and Developmental Disorders

DDST: Denver Developmental Screening Test

DSM-IV-TR: International Classification of Mental Disorders of the American Association of Psychiatry

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FAECAP: Spanish Federation of Community Nursing and Primary Care

FEAPS: Spanish Confederation of Organisations in favour of People with Intellectual Disabilities

FESPAU: Spanish Federation of Associations of Parents of Autistic Children

FISH: fluorescent in situ hybridisation (diagnostic test)

ICD-9, 10: International Classification of Diseases, version 9 and 10.

INAHTA: International Network of Agencies for Health Technology Assessment (International Network of Agencies for Health Technology Assessment)

INE: National Statistics Institute

MC: monozygotic (twin)

M-CHAT: Modified Checklist for Autism in Toddlers

M-CHAT: Modified Checklist for Autism in Toddlers

MLPA: Multiplex ligation-dependent probe amplification techniques (diagnostic test)

NANDA: Nursing Diagnoses according to the North American Nursing Diagnosis Association

NEA: Spanish Association of Neuropsychiatry

NHS: National Health Service

NICE: National Institute for Clinical Excellence

PC: Primary Care

PDD: Pervasive Developmental Disorders

PEDS: Parents' Evaluation of Development Status

PEDS: Parent's Evaluation Scale or Developmental Status

PICO: Patient / Intervention / Comparison / Outcome or Result

SCQ: Social Communication Questionnaire

SEMERGEN: Spanish Society of Primary Care Physicians

SEMFYC: Spanish Society for Family and Community Medicine

SENP: Spanish Society for Paediatric Neurology

SEP: Spanish Society of Psychiatry

SIGN: Scottish Intercollegiate Guidelines Network

SP: Specialised Care

SW: West Syndrome

TGD-NOS: Pervasive Developmental disorders not otherwise specified

TV: TV

UETS: Unit for Health Technology Assessment

WHO: World Health Organisation

WONCA: World Organisation of National Colleges, Academies and Academia Associations of General Practitioners / Family Physicians (World Organisation of National Universities, Academies and Academic Association of General Practitioners and Family)

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Appendix 11. Declaration of interest

A conflict of interest occurs in circumstances where the professional opinion on a primary interest, such as patient safety or validity of the investigation, may be influenced too much by other secondary interest, be it due to financial benefit, prestige and personal or professional promotion.

In the relationship between professionals and the healthcare industry (pharmaceutical, medical technology, etc.) various types of financial interactions can be considered.

- Support and funding of research
- Be employed as a consultant for a pharmaceutical company
- Be a shareholder or financial interest in a pharmaceutical company

In turn, these potential conflicts of interest in the development of CPGs are considered of two types:

– Personal interests: involve fees or personal benefits to a member of the team.

– Non-personal income: involves funding that benefits the department or unit under the direct responsibility of a member of the team, even if this person does not receive it personally. Financial assistance to create a unit or department, financial support for the recruitment in these units or research funding in the unit can be considered non-personal benefits.

The potential conflict of interest exists irrespective of whether the practitioner believes that such relationships have or not influence on his scientific criteria. Attached is a form of the declaration of interest conflict designed to reflect the points stated above.

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Interest Statement Form

- Full Name:

- Profession:

- Institution where he / she works (including address):

- Institution represented (association, scientific institution, etc., answer only if different from the institution stated above):

- Telephone: In the morning:

In the afternoon:

- E-mail to send documentation (@):

Mr / Ms _____ with ID _____, having read the policy-making Programme of the Clinical Practice Guidelines (CPG) based on evidence, in order to help on clinical decision making in the National Health System (NHS) declare to have

YES NO

conflict of interest related to activities in connection with the subject of the CPG for the past three years.

If yes, please specify which in Annexes I to III.

Signature: Date:

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Annex 1. Personal Interests

	Activity	Institution	Date
Funding to participate in an investigation			
Consulting for a pharmaceutical company / other technologies			
Shareholder / commercial interests in a company (patents)			
Economic interests in a private company related to the health sector (as owner, employee, shareholder, private consultation...), which may be significant in relation to the authorship of the guide			
Conflicts of interest of non-economic nature that can be significant in relation to the authorship of the guide			

Annex 2. Non- Personal Interests

	Activity	Institution	Date
Funding or financial assistance to set up a unit or service			
Provide significant material to the unit or services			
Shareholder / commercial interests in a company (patents)			
Procurement or financial assistance to recruit staff on the unit or service			
Financial assistance to fund an investigation			

Annex 3. Other possible conflicts of interest not mentioned in the previous tables (please specify)

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