Children and adolescents with developmental disabilities in the pediatric outpatient clinic

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Abstract

Objective: To review the literature on outpatient care of children and adolescents with developmental disabilities, focusing on prevention, early diagnosis, treatment, outcomes and rehabilitation.

Sources: Search of Medline and Lilacs databases; publications of scientific committees of institutions for children with special needs; and outpatient care reports of reference centers for the treatment of children and adolescents with mental and developmental disabilities.

Summary of the findings: This population presents health problems associated with the basic pathology and its consequences in addition to the usual problems of the specific age group. The etiology of developmental disorders and the main characteristics of each type of disability are discussed. Special attention is given to outpatient pediatric care.

Conclusions: The most recent study of the Brazilian population, performed in 2000, showed that 14.5% of this population presented some type of developmental deficiency. Therefore, developmental problems are one of the most prevalent health problems among children and adolescents. Consequently, pediatricians need to be prepared to evaluate and identify factors that may influence normal children development. Pediatricians are responsible for prevention, early diagnosis and coordination of the multidisciplinary treatment of these patients in addition to basic medical assistance, which is essential to guarantee the patients’ good quality of life.


Introduction

According to data from the World Health Organization (WHO), in peacetime, at least 10% of children of any country are born or acquire physical, mental or sensory disabilities that will interfere with their development.1

In our country, the 2000 census shows a prevalence of approximately 24.5 million people with disabilities, which accounts for 14.5% of Brazil’s population.2 However,
according to a document issued by the Ministry of Health in 1991, only 2% of these individuals received some kind of assistance, either from the private or public sector, with insignificant improvement of this situation.³

This situation is rooted in the historical process of public policies regarding health care and rehabilitation of these individuals, which has been characterized by the passing through of financial resources to philanthropic organizations over the years, keeping the attention towards disabilities within the realms of assistentialism and humanitarism.³

Looking to the figures shown above, there exists a higher prevalence of disabilities among children and adolescents than any other kind of pathology; however, they are not included in treatment protocols at most general pediatric outpatient clinics.

Children and adolescents with developmental disabilities show different characteristics as to the need of assistance, according to the underlying disease and to the limitations imposed by it, and are amenable to all types of infections and general disorders that are characteristic of childhood and adolescence.

Therefore, it is necessary that the pediatrician be prepared to include patients who require special attention in his/her routine practice. This article touches upon aspects that relate better to the neuropsychomotor development of the child and its dysfunctions, as well as to the major characteristics of children and adolescents with disabilities and to their special needs, with the aim of defining the role of the pediatrician in the prevention and early identification of developmental disabilities and in the health assistance these patients are provided with.

**Definition of development**

To define development is not an easy task, in view of the theories and aspects that can be adopted. For the pediatrician, there is the classic definition by Marcondes et al. (1991),⁴ which states that “development is the increased capacity of an individual to perform more complex tasks;” on the other hand, pediatric neurologists always look to the maturation of the nervous central system; likewise, a psychologist will think about the cognitive aspects, intelligence, interaction with the environment, whereas a psychoanalyst will give more importance to relationships and to the formation of psychism.⁵

In a broader sense, child development is a process that begins in utero and encompasses several aspects, such as physical growth, neurological maturation and construction of abilities related to behavior, and to the child’s cognitive, social and emotional spheres. The aim is to render the child able to meet his/her requirements and those of the environment, by taking into account his/her life context.

**Risk factors for developmental disabilities**

Risk factors are defined as a series of biological or environmental conditions that increase the probability of neuropsychomotor development deficits in children.⁶ This definition, albeit didactic, is not always easily used in everyday medical practice, since under several circumstances biological and environmental factors overlap, increasing the probability of injuries.⁶⁹ Some authors separate the biological risk from established ones (defined medical disorders) such as inborn errors of metabolism, congenital malformations, Down’s syndrome and other genetic syndromes.¹⁰

Environmental risks, related to the general and family environment in which children and adolescents live, are characterized by omission or action, such as poor health assistance, lack of social and educational resources (or lack of interest in providing them), lack of policies and measures for accident prevention, violence and others.⁷⁻⁹ They are partly related to poverty, but do not solely depend upon it, since many of them such as accidents, negligence and other forms of domestic violence occur in all socioeconomic classes (Table 1).

Children at higher risk, that is, those weighing less than 1,500 g at birth and with significant neonatal intercurrent diseases, should be followed up by a multidisciplinary team at referral centers, whenever possible, with the aim of detecting and early treating any sign of developmental disability. Some recommendations should be followed when treating these children:

- document the whole investigation, diagnoses and treatments used and give the family a copy of these documents. This avoids having to repeat exams and can guide other pediatricians in a future follow-up;
- tell the family of the importance of a differentiated follow-up;
- refer the patient to other assessments and specialists, if necessary, in time for treatment;
- promote mother-baby interaction, encouraging mothers to hold, touch, caress and talk to the baby, even at ICUs and also in the cases of severely ill babies;
- the functional diagnosis is sufficient for referring the patient to early intervention;
- always be attentive to development and do not hesitate to ask for other specialists’ or professionals’ help when you observe delays or deficits.

**Early detection of developmental disabilities**

Early identification and intervention are essential for the prognosis of children with developmental disabilities, which makes the assessment of this process indispensable in every appointment with a pediatrician.¹¹

In general, the more severe the developmental disability in a child, the earlier the identification by the
Data on possible risk factors for developmental disabilities should also be obtained, from conception to the moment the child acquires abilities. The opinion of family members about the growth of the child should also be considered. There is common agreement in literature that parents are good observers and precise detectors of disabilities in their children, showing high sensitivity, specificity and predictive value of their opinion for the detection of developmental disorders.13

The general physical examination and the neurological assessment, sensory evaluation, and assessment of children’s acquisitions are the basis for evaluating child growth and development.11

It is important to offer infants an object they feel attracted to so that their interest, coordination and interaction with the mother and examiner can be evaluated. In older children, speech, lucidity and location in time and space should be assessed. Pencil and paper are important ancillary tools, as drawings or basic writing...
activities may provide relevant data on comprehension, abilities and motor function.\textsuperscript{11,12}

On physical examination, some characteristics may suggest diseases that are concurrent with developmental disabilities. In this case, special attention should be given to weight, height and head circumference parameters; presence of congenital diseases and skin disorders, since the association of developmental disabilities with neurocutaneous syndromes is quite common. The palpation of the abdominal region in search of visceromegalies, frequently observed in several developmental disorders, should be performed.\textsuperscript{11}

The assessment of the sensory system, especially of hearing and sight, should be made from the first months of life. This assessment will be seen in detail when the characteristics of each deficiency are analyzed.

The evaluation of the child’s abilities plays a crucial role in the diagnostic process. The literature presents several development scales, most of which are based on Gesell development schedules. These scales are valuable in the evaluation process as a way to systematize the examination and, in our opinion, only during the first years of life, stage at which the biological and maturational aspect predominates in the development process. As they are based on populations of extremely different cultural contexts, they can hardly be applied to older children. In addition, the scales usually analyze what the child does, but are not capable to envisage development as a process or to consider the child’s potential.\textsuperscript{12}

**Major clinical signs**

Disabilities are classically divided into mental, physical, sensory (auditory and visual) and mixed, in which all types of deficits and limitations may be present.\textsuperscript{1} In this classification, we found it important to group patients with pervasive developmental disabilities.

**Special children and adolescents and their special needs**

When dealing with a child or adolescent with special needs, some management procedures may hinder the relationship between patient, family and health professionals, delay the measures necessary for diagnosis, habilitation or rehabilitation and, sometimes, cause disastrous effects on the prognosis of these patients.

It is not necessary to have a specialty in order to follow up development, detect signs of developmental disorders or meet the basic health requirements of a patient with these disabilities, since the signs of childhood diseases, and viral or bacterial infections and the treatment will almost always be the same as those of other children.

According to the type of developmental disability, basic rules for treatment should be followed so that a better prognosis and adherence of the patient and the family to the proposed treatment can be obtained (Tables 2 through 6).\textsuperscript{14}

**Specific characteristics of developmental disabilities**

**When mental retardation predominates**

Approximately 50% of those with special needs have mental retardation. According to the American Association of Mental Retardation (AAMR)\textsuperscript{15} and to the Diagnostic and Statistical Manual of Mental Disorders

<table>
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<tr>
<th>Table 2 - Guidelines for management of children or adolescents with disabilities</th>
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<tbody>
<tr>
<td>1. Do not present a definite etiologic diagnosis within a short period of time. Assess your patient, establish bonds, consider the possibilities of a developmental problem that requires further investigation and other professionals’ advice.</td>
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<td>2. The functional diagnosis should be established in the first assessment in order to allow referral to treatment with a multidisciplinary team and to avoid further delays and sequelae.</td>
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<td>3. The prognosis should be released only when the complete assessment is finished by the multidisciplinary team, and after the family is considered ready to accept and treat the problem.</td>
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<td>4. Do not make predictions regarding the prognosis. Avoid saying sentences such as “he’s not going to walk” “He’s not going to learn or listen...”. These predictions might become reality, since the family abandons the treatment. The developing child receiving appropriate treatment is able to achieve a surprising recovery, since there are several manners to see, listen, speak and live in a society...</td>
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<td>5. Always refer your patient to interdisciplinary treatment with specialized care for the areas that present or might present disabilities.</td>
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<td>6. Emotional support is extremely important in the beginning of the disability management and in several other phases of the child and adolescent’s development.</td>
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<td>7. Avoid repetition of the examinations or investigations that do not aim at the improvement of assessment and treatment, which will only cause suffering and increase everyone’s frustrations when they do not have as a consequence new options of treatment or cure.</td>
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<td>8. Disabled patients have special sensitivity when it comes to understanding environment and people. They are able to perceive the professional’s lack of interest, even in the most severe cases of disability.</td>
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<td>9. The first one to receive advice regarding the treatment should be the patient, always respecting his/her ability to understand and using language that he/she is able to understand. Then the guardians should be advised about the treatment and its importance, since the treatment compliance and follow-up depends on them.</td>
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<td>10. Try to have a record of the addresses of support groups and associations of disability carriers in your city. These associations usually offer special services to these families regarding the acceptance of the disability, and many of them provide excellent early management and rehabilitation services.</td>
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mental retardation is defined as the remarkable reduction of intellectual functions to a level that is significantly lower than the average, initiated during the developmental stage and associated with limitations in at least two aspects of adaptive functions: communication, self-care, daily activities, social abilities, use of community resources, autonomy, school aptitude, leisure and work.

This definition eliminated the classification of mental retardation as mild, moderate, severe or profound, and admits that the level of development of an individual depends not only on the severity of mental involvement, but also on the patient’s life history, family and social support and on existing opportunities.

Perinatal hypoxia and congenital infections are the major causes of mental retardation, as well as Down’s syndrome, which is the most frequent genetic chromosomal syndrome. In boys, one should not forget the fragile X syndrome. The Fetal Alcohol Syndrome should be suspected in all children with low weight/height gain, microcephaly, hypoplastic philtrum, thin upper lip, retrognathia, and short palpebral fissures. Among chronic encephalopathies in children, inborn errors of metabolism and the acquired immunodeficiency syndrome are noteworthy.

Basic care procedures for individuals with mental retardation

Individuals with mental retardation are often enthusiastic, joyful, caring and communicative. Their sensitivity is sharp and their cleverness in assessing the environment and the people who surround them is out of the ordinary. Table 3 presents some important measures for the care of such patients.

Table 3 - Basic guidelines for management of mentally disabled patients

1. To act naturally and use affectionate words is the best way to make the patients to establish bonds, allow the clinical examination and accept the treatment.
2. Over protection should always be avoided.
3. Each procedure or approach should be thoroughly explained and they should not be handled as a schedule handed in at the patient’s arrival.
4. These patients need more time to understand why they have to go through physical examinations so that they can feel safe.
5. Be aware that the mentally disabled patient does not have a mental disease. He/she presents a consequence of a disease or lesion - it is a “condition of being”.
6. Treat the patients like children if they are children and like adolescents if they are adolescents, always respecting their sexuality and advising their families to do the same.
7. Always consider if the current clinical status is part of the original pathological evolution, or if it has a different origin and deserves to be further investigated and receive new treatment.

Children with developmental disabilities - Miranda LP et alii
Psychiatric disorders: emotional disorders, especially depression and Alzheimer’s disease have been frequently described in young individuals and adults.

When physical deficiency predominate

In the first years of life, cerebral palsy is the most common physical deficiency. The term cerebral palsy, albeit widely used, was employed to define a series of different diseases, which not always involve palsy, and which often are of noncerebral origin. Cerebral palsy is one of the forms of chronic encephalopathy in children. Therefore, all progressive disorders are ruled out. It is characterized by motor disorders due to interference with the development of the central nervous system, possibly causing voluntary motion disorders, muscle tone disorders, hyperkinesis, usually concomitant with learning, communication or speech disorders. Individuals with cerebral palsy may have normal intelligence quotient and the severity of the motor disorder not always corresponds to the same cognitive involvement.  

Children with cerebral palsy often have some particular characteristics that should be known by the pediatrician, such as:

- global developmental disabilities, with necessity of systematic assessment of hearing and sight;
- swallowing difficulty and, later on, chewing difficulty;
- gastroesophageal reflux and cricopharyngeal incoordination;
- malnutrition and eating disorders caused by swallowing difficulty and recurrent infections;
- recurrent respiratory diseases, either due to hypotonia of respiratory muscles, or due to remaining in the supine position for a long time or due to aspiration;
- intestinal constipation produced by hypotonia and eating disorders;
- neurogenic bladder, for which urological assessment should be performed at and after the third year of age.

Constant spasticity leads to postural disorders, such as flexion of upper limbs and hyperextension with abduction of lower limbs, in an X position, which may cause shortening of muscles and tendons, joint immobilizations and usually painful complications (luxation or subluxation of the hip joint).

When sensory disorder predominates

Hearing deficiency

The WHO estimates that 15% of individuals with disabilities have hearing disorders. The incidence of hearing loss in healthy newborns is estimated to range from one to three newborns per every 1,000 live births and from two to four per every 100 newborns that require intensive care. This way, hearing assessment should be made in all newborn infants, is essential and should be early carried out up to the third month of life in those at risk or at the NICU, so that intervention occurs before the sixth month. Intervention up to this age significantly improves the possibilities of language acquisition. Lack of stimulus may increase sequelae at the cortical level, which will impair hearing ability for life, even if peripheral problems are later treated.

Neonatal risk factors for hearing deficiency are similar to those of general disabilities, but also include family history of congenital hearing deficiency, consanguinity, craniofacial anomalies, genetic syndromes and use of ototoxic drugs. Bacterial meningitis, recurrent otitis media, head traumas and neurodegenerative disorders are important causes after the neonatal period. In our setting, cases secondary to congenital rubella are unfortunately quite prevalent.

<table>
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<tr>
<th>Table 4 - Basic guidelines for management of patients with physical disabilities</th>
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<tr>
<td>1. Be careful when approaching these patients and when making</td>
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<td>comments and presenting prognosis to the parents while the</td>
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<td>patient is present. The existence of physical disabilities</td>
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<td>does not necessarily mean there are cognitive deficiencies.</td>
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<td>2. Always talk directly to the patient. Be patient and respect</td>
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<td>your patient’s pace. Besides the walking problems, they often</td>
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<tr>
<td>might present speaking problems and difficulties to control the</td>
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<td>movements. These difficulties are usually increased by fear</td>
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<td>and lack of confidence.</td>
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<td>3. Usual pathologies in childhood should be treated by the</td>
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<td>same procedures employed with other patients. The history of</td>
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<td>infections and previous and current treatments should be</td>
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<td>studied in order to assess interaction, contraindication or</td>
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<td>resistance to some medications.</td>
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<td>4. When there is diagnosis of malnutrition, assess the level</td>
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<td>of malnutrition and verify the possibility of malnutrition</td>
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<td>being a consequence of the basic disease or secondary to</td>
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<td>negligence. The treatment and the approach to the parents will</td>
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<td>be completely different in each case.</td>
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<td>5. Regarding cases of tetraplegia or severe cerebral palsy,</td>
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<td>advise your patient about appropriate feeding position in</td>
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<td>order to avoid hyperextension of the head, which increases</td>
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<td>the occurrence of bronchial aspiration.</td>
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<td>6. Always refer your patient to interdisciplinary follow-up</td>
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<td>in order to avoid severe physical and mental sequelae, and</td>
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<td>family dysfunction.</td>
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<td>7. If the child or adolescent uses a wheelchair, do not</td>
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<tr>
<td>touch or handle it without your patient’s permission, since</td>
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<td>the constant use and the dependency on the wheelchair makes</td>
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<td>the patient to consider it as part of his/her own body. If</td>
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<tr>
<td>the patient uses crutches or other support system, always</td>
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<td>make sure the patients can easily reach them.</td>
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<tr>
<td>8. Always keep good accessibility conditions and require them</td>
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<td>in public health care centers. The inclusion of the disabled</td>
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<tr>
<td>patient in general pediatric outpatient clinics depends</td>
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<td>mostly on the pediatrician, and also on good accessibility to</td>
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<td>health care centers, without constraints or limitations</td>
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<td>imposed by the physical environment.</td>
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Hearing deficiencies should be suspected in all children with absence of reaction to sounds, delayed language acquisition, preference for loud sounds, and constant distraction and inattentiveness. The attitudes of infants with hearing impairment are often mistaken for autistic behavior and, in older children, they are mistaken for mental retardation.24,27

Visual impairment
Every child in his/her first months of life should be submitted to systematized ophthalmologic assessment. During this period, the evaluation of the red and photomotor reflex, fixation and following of objects is essential. Those children with altered reflexes, leukocoria, delayed fixation or following of objects, different-sized eyeballs, constant blinking, photophobia, nystagmus, fixed eye deviation and alternating deviation after the fifth month of life must be evaluated.30

Retinopathy of prematurity should be investigated in every child with birthweight less than 1,500g, gestational age less than or equal to 28 weeks or in those weighing between 1,500g and 2,000g with clinical instability and in newborns that required oxygen therapy at high concentrations or for a prolonged time. These children should be submitted to at least two examinations of the ocular fundus, the first of which should be performed between the fourth and sixth weeks of life. This examination should be carried out by a qualified professional, with a binocular ophthalmoscope for accurate and early diagnosis.31

Basic care regarding patients with visual impairment
Patients with visual impairment develop a more pronounced auditory and tactile perception than the general population. Therefore, some management measures are necessary (Table 6).

Table 5 - Guidelines for management of children and adolescents with hearing deficiency

| 1. | Find a way to communicate. Ask the parents how they communicate with their child and try to do the same! While speaking, always stand in front of your patient, pronounce the words in a clear manner, do not speak fast, make sure that your patient is able to see your face. |
| 2. | Always explain, through gesture and signs, each procedure during physical examination. Make sure that the child sees what is going on. |
| 3. | Use a normal voice tone, unless the patient asks you to speak louder. |
| 4. | If your patient tries to talk to you and you do not understand, asks him/her to repeat or to write down. Even if you need someone to help you to communicate, talk directly to the child or adolescent. |
| 5. | The presence of the guardians during examination is essential to make the child feel safe. |

Table 6 - Guidelines for management of patients with visual impairment

| 1. | There is no need to use a loud voice tone when talking to patients with visual impairment. These patients usually have better hearing abilities than people in general. |
| 2. | When performing physical examination, explain the procedure before performing it and warn the patient about everything you are going to do before touching him/her. |
| 3. | When dealing with an older child or an adolescent, in the presence of the parents, ask the patient if he/she wishes to touch your face to know how you look like. Through the voice tone, the way someone speaks and behaves, the patient with visual impairment is able to create an image of the person who is in front of him/her, and often this image is quite similar to the real one. |
| 4. | Never use medication that may increase the intraocular pressure without appropriate diagnosis of the visual pathology, and without discussing the use of the medication with his/her ophthalmologist. |
When relationship disorders predominate

The construction of subjectivity is only possible through the interaction between the child, his/her caregivers, and collective and institutional instances in which the child is inserted. So that the child can develop as an individual, it is important that there exists a place for him/her in the imagination of others, in which the mutual relationship between the child and his/her mother or substitute plays a vital role. Therefore, it is important to observe the relationship between the child and his/her mother or caregiver.

A look, a smile and estrangement are important markers of emotional and relational development.

Pervasive developmental disabilities encompass a wide range of disorders that begin in infancy and are characterized by changes in social interaction, communication, imaginative activity and a small repertoire of activities of interest. Some of these children have cognitive disorders, of which autism is the most serious. The diagnosis of autism may be established in the first months of life by means of flaws in emotional development. These children usually develop their motor functions normally but present extensive speech involvement.

Children, adolescents and their families in relation to the disability

Most children who are treated at the pediatric outpatient clinic present with developmental disabilities secondary to predisposing factors during the prenatal, perinatal or neonatal period. Thus, these disabilities occur very early on and the child’s relationship with the world is difficult and limited at the beginning.

In older children and adolescents, external, infectious and degenerative causes are involved. In this case, the plight is not only of the family but also of the child, who needs to adapt to the limitations imposed to a formerly healthy body. The patient’s plight is more intense due to revolt, depression and even willingness to die.

A developmental disability provokes a series of reactions, depending on the way this diagnosis and prognosis is presented and on the support the family receives especially in the first moment, from the health team.

The initial reaction to the diagnosis of risk or disability is one of doubt or denial - “there must be some mistake; I haven’t made anything wrong to deserve this.” Consequently, other professionals are sought and new exams are requested with an attempt to find another diagnosis or possibility of cure.

When the diagnosis is reaffirmed, a feeling of guilt appears - “what have I done wrong? What have I done to deserve this?” These are feelings recalled from the memory of a lifetime in search of a reason for the loss of the dream of having a healthy child or the loss of the “normal” body in case of acquired disabilities.

A feeling of revolt follows - “Why me?” - in this case, everything and everyone is to blame, and pediatricians should therefore be ready to recognize this phase, since it involves extreme aggressiveness of parents and patient towards health care professions. - “Where have doctors and the others failed me and my child?”

After revolt, when denying, blaming or fighting against the problem is no longer possible, comes the need to face reality and, as a consequence, sadness, the plight of loss, which may develop into depression and hinder adherence to any treatment. After this plight comes acceptance, which does not always occur, and afterwards, recovery. Many families or disabled individuals insist on some of these phases, such as that of revolt or blame, always trying to have new exams done, have new diagnoses made or even trying to find a miraculous cure, and never fully adhere to the proposed treatment.

It is important to know in which phase the family and patient are and to respect this. The problem should not be dealt with on a personal basis, since this phase is natural and necessary for recovery (the last phase) to occur. The last phase is the search for what it is possible to do, with adherence to the proposed treatments, habilitation and rehabilitation, so that children or adolescents can accept themselves and be accepted by the family as they are, with some difficulty, of course, but also with the possibility that they can compensate for or even overcome their limitations and maintain a good quality of life if they have proper support.

Treatment and rehabilitation

Except for some inborn errors of metabolism and congenital infections, no single and specific treatment is available for developmental disabilities.

New knowledge about brain plasticity in humans reiterates that health professionals should intervene early on and should not prophesize prognoses for these patients. Stimulation in the first three years of life for children with delayed development or for those at risk improves their performance; the earlier the stimulation, the better.

Rehabilitation is the process whereby the disabled individual can reach an optimal physical, mental and/or functional social condition, and thus be able to change his own life. The whole rehabilitation should focus on the abilities of the individual, whose integrity and dignity should be respected. When planning rehabilitation and support programs, it is essential that the routine, possibilities and structures of the family and community be considered, enhancing the response to the needs of a person with developmental disabilities.

Within this context, rehabilitation programs should be incorporated in a decentralized way to the network of health services and should include not only a multidisciplinary approach, but also the participation of the community and family.
Inclusion

The inclusion of these children and adolescents in the community is fundamental for their treatment. Inclusion means giving them the same opportunities the general population is given. It is important that society’s general system - physical and cultural environment, housing, transportation, social and health services, education and job opportunities, cultural and social life - be available to all. In general, what determines the effect of a certain disability in someone’s life is his or her experience with the environment. Therefore, public policies targeted at the rehabilitation of individuals with developmental disabilities are not enough. It is necessary to offer equal participation in various aspects of social life.

Our conclusion is that when we say these children and adolescents have rights, we do not only mean the rights involving their peculiarities, but also their rights to participation in society, with their differences and singularity.

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