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Follow up of the children seen at the "Ramón Sardá" maternal hospital of Buenos Aires, Argentina

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Abstract

We show the results of the follow-up program for children with Down Syndrome, seeing by the ATIENDO group in the Maternal Hospital Ramón Sardá the Buenos Aires Argentina. We describe the program, the study population, associated illness, the results, and intervention approaches to promote integration of these children in the family and society.

Keywords: Down Syndrome. Interdisciplinary follow up.

1. Introduction

Down syndrome (DS) is the most frequent chromosome alteration in born-alive newborns, with an incidence of 1 per 700 to 1,000 births, irrespective of race, geography, or social class. There is a distinctive associated phenotype, as well as developmental, structural and functional impairments to a number of systems and organs, most significantly some degree of learning disability, a greater risk of congenital heart defects, digestive tract malformation, sensory impairment (sight and hearing), and thyroid disease, all of which require long-term follow-up (1). It is worth noting that a child either has DS or does not – there is no spectrum (2).

The most common cause is the presence of an extra 21st chromosome, occurring in 95% of cases. In over 80%, the trisomy is of maternal origin; 4% are caused by a chromosome translocation and 1-3% have a mosaic trisomy.

Quality of life and life expectancy in DS children have increased dramatically in the last few years. Considerable personal independence and integration into society are now possible, which makes early support for parents and raising their awareness of the importance of early stimulation (ES) crucial for these children to be able to reach their full potential and development in life (3).

Why a Follow-up Program?

Multiple risk factors compound this disability. The effects of an illness or congenital disorder can be heightened by unsuitable surroundings which can endanger and delay development, leading to a population at risk due to mixed environmental and biological causes (3).

a. Environmental risk: In the early stages many children spend a large part of the day stuck in a crib, cot or stroller where they are unable to practice their motor skills. The lack of space and mobility restricts their exploratory behavior. Although their environment contains sensory stimuli (for touch, sight, hearing, smell, etc.), these are disorganized and hazardous for young children, with an anarchic reinforcement system and no planning or care to avoid damage from external causes. The parents, barely able to keep their heads above water, struggle to support each other emotionally. In financially unstable families this bond is jeopardized and becomes more difficult, with the TV often serving as the child's only companion. This is compounded by the high rate of marital breakdown and subsequent separation following the birth of a child with a genetic disorder.

b. Biological risk. Due to the genetic disorder.

Vulnerability can be curtailed by means of the protection and resilience factors (emotional, cognitive, environmental and other resources) (4,5,6) listed below:

1. Coping strategies (concrete context-based processes, varying widely depending on triggering conditions). *2.* Early, prolonged and continuous *baby-mother-father contact 3. Skin-to-skin contact. 4. Breastfeeding. 5. Visits from siblings and grandparents* for babies in neonatal intensive care units. *6. Fundamental organization and limits* in everyday life. *7. Support available locally*: grandparents, friends and neighbors. *8. Support networks of families* who have gone through the same experience, parent groups, and national and international organizations. *9. Support from the healthcare team.*

The importance of interdisciplinary work in following up children with DS

Child development and growth must be seen as a total, gradual process encompassing a range of factors that must be brought together. A team of professionals who complement each other is assigned to the child and family to cover all aspects of development, providing a broad, cross-cutting perspective. The pediatrician acts as coordinator and liaises with the different specialists, rather like a GP (6, 7).

What is the 'Atiendo´ Group?

Over the course of their life, some children with DS may suffer health problems caused by complications that could have been avoided by providing follow-up to ensure prevention and early treatment. At the Ramón Sardá Maternity Hospital (HMIRS), routine follow-up of children with DS was set up some years ago through a total birth-to-school-age healthcare program. Its aim is to enhance children's quality of life and foster their integration into society, helping parents to achieve this by seeing all individual success as based on ability rather than disability (8, 9). The 'At.i.e.n.do' Group carries out a range of activities:

1) Informing families. Before even being conceived, children exist in their parents' plans and desires; pregnancy involves imagining the child and communicating with him or her ('talking to baby'). The baby represents the continuity of their parents' wishes and they need to recognize themselves in her. When a child is born with DS, the image of the complete, normal, 'idealized' baby is shattered. On finding out that their child is a carrier of an anomaly, the parents are disappointed and react according to their expectations and personality. The baby that has been born is not what they anticipated, and their grief is twofold: for the child of their fantasies, who has died, and the presence in her place of an unexpected child whom they struggle to love. The news must be conveyed by professionals trained to handle this type of situation and emotionally able to deal with the parents' anxiety, to 'sustain' this baby, and to field questions that for the parents have no satisfactory answers. Faced with these reactions and queries, healthcare staff help the parents to accept reality, guiding them as they learn to live with their child and give her love, acceptance and belonging. DS is not portrayed as an illness; the child is posited as a subject, which makes it easier to achieve inclusion in family life. This helps the parents to process the disability and bond with their real child, seeing beyond the phenotype to her individuality and similarities with other members of the family, in a twofold initial identification that will underpin the child's filiation, the things that make her unique in the world, and her place in the family dynamic. Receiving such devastating news is extremely painful and there are no prescriptions, rules or magic recipes on how to give it. In Ancient Greece, the bringer of bad tidings was killed. The information is given in the delivery room in the presence of the pediatrician, psychologist and geneticist.

2) Total healthcare of children with DS from birth in the hospital setting. Meeting the needs of the children and their families. Taking care of health, prevention, growth and development, in addition to associated illnesses. Taking on the responsibility of physical, mental and emotional progress. Dealing with social and environmental factors that have as much of a bearing on the child's welfare as organs and biological processes.

3) Early Stimulation (ES) Program. What is Early Stimulation? ES is a discipline aimed at treating babies and young children who have or may have developmental impairments (4). Parents use sessions to discuss their support needs and fears, and recognize and get to know their 'real' baby and her possibilities. This is a space where parents can accompany their baby's development so that they can discover their place as parents, include this child in their family life, and deal with mixed feelings (10, 11). In Argentina, prejudice and taboo regarding Down syndrome is rife and it is therefore vital to show that children who receive ES from birth perform better in all areas of development (4). The child's progress encourages mom and dad to help achieve integration in their surroundings and, in turn, to act as agents of change in the rest of society, depending on their family, social/economic and cultural status. ES takes place once a week at the hospital; when this is not possible, the child is referred to a centre near home.

4) Early Educational Psychology Socialization Group (EEP). Parents discover that social integration and insertion begin before a child starts school; it is in fact the step prior to enrolment in a special or mainstream kindergarten, according to individual needs. ES gives way to a new stage: EEP, which is a progressive journey encompassing changes in counselor, approach and the stance taken by parents

and child up until school enrolment, to facilitate their first social experience outside the family. These early shared experiences will allow the children to strike relationships with space, objects, and other people. In terms of emotions, they will help progressively to build up self-esteem and lay solid foundations for learning, the bedrock for future cognitive acquisitions (10, 11). The children are taught basic social skills (cooperation and reciprocity) for interacting with others.

When the child is ready, at around two years old, ES sessions finish and the socialization group begins. Meetings are held weekly with groups of 3 to 4 children and aim to prepare them for starting school.

5) Parents meetings. This is not a self-help group but a group run for counseling, solidarity and support of and between families, aimed at helping parents to cope in everyday life with their children. Parents are the rock that supports a child through their day-to-day actions, stimulation, and performance of parental roles, so that the child can grow up to be independent. The parents are proactive participants, spokespersons for the patterns acquired in primary health prevention who circulate the booklets given out on the issues covered. These meetings foster the family's relationship with the child, promote breastfeeding and enable parents to meet other parents of children with DS. The sessions deal with the demands, concerns and hurdles that arise at each stage of the child's life and group outcomes are construed as a means of support and guidance. The queries and proposals raised by some parents are shared by the majority. Specific issues are dealt with in individual therapy sessions. The core topics are presented by experts in each area, and 'trigger' questions are used to develop related subjects, with close relatives and friends invited to share. This is a space where they can: **a)** Freely express their feelings, opinions and proposals. **b)** Tackle concerns, myths, fears and prejudices on DS. **c)** Work on their position as parents and the place granted to their child. **d)** Discover the characteristics of each child through the diversity of the dynamic. **e)** Uncover the path towards the child's creation of a sense of self and healthy development. **f)** Discover with their child the pleasure of growing up and becoming someone who can think, wish, play and be happy.

6) Activities to foster the best development possible and social integration.

7) ES referral network for centers near home, if not taken at the HMIRS.

8) Guidance on integration at school.

9) Welfare benefits advice: higher family allowance for parents who are employees, disability allowance, free public transport pass, and various types of grants (food hampers, dietary support plan, etc.).

2. Objectives

a. General objective

– Long-term interdisciplinary care of children with DS and their families

b. Specific objectives

– Identify the population of children with DS

– Detect and treat associated conditions as early as possible

– Assess growth in children from 0 to 3 years

– Provide a space where parents in the same situation can meet and find support

– Foster the best development of each child's capacities through ES

– Offer the first social experiences outside the home (before mainstream or special kindergarten, according to each child's needs (EEP))

3. Material and method

A prospective, longitudinal and descriptive study was undertaken of 107 children with DS, mostly born at the HMIRS (91%: 98/107) and in other health centers (8.4%: 9/107) who voluntarily attend follow-up for three years after birth. This work is still underway and the data presented here constitutes a first step in this care and research. Cognitive level is currently assessed using the Argentinean Sensory-Motor Intelligence Scale ('Escala Argentina de Inteligencia Sensorio Motor ('Argentinean Sensory-Motor Intelligence Scale') (12) and psychomotor development using the Escala de Evaluación del Desarrollo Psicomotor ('Psychomotor Development Assessment Scale') (13). The results shall be analyzed over the course of this year.

- **Population:** all children with DS born at the HMIRS between 1/1/1996 and 6/30/2002, and 9 children born at other centers, monitored at the 'At.i.e.n.do' Group until the age of three.
- **Drop-out rate:** before the age of 2 this is 7.4%, a lower rate than other groups followed at the HMIRS (14). Between 2 and 3 years the drop-out rate is 14%, most of whom are children not born at the HMIRS.
- **Methodology:**

Growth assessment based on US growth charts for children with DS (15).

Check-ups set out in follow-up protocol (cardiology, genetics, speech therapy, ophthalmology, thyroid assessment, etc.) (16, 17, 18).

- **Mode of work**. **Includes:**

1. Informing families: The genetics team handles the clinical examination to confirm the diagnosis (phenotype) and performs the karyotyping and genetic assessment. Parents are told how DS occurs and what the risks are for future pregnancies, according to

whether they are healthy or carriers of a translocation.

The initial information is given during the hospital stay together with the psychology service.

2. Total child care: Care is provided for healthy children as well as for those with intercurrent conditions from birth to 3 years. In the child's first year the check-ups are monthly, in the second they are quarterly and henceforth they are six-monthly. The HMIRS Pediatric Outpatient Department runs these as a Day Hospital service (all tests, assessments and interdepartmental consultations are done on the same day): **a)** growth assessment; **b)** breastfeeding promotion and support (the HMIRS has been designated a Mother-Child Friendly Hospital as part of the UNICEF initiative); **c)** guidance on baby care; **d)** health education; **e)** health prevention and promotion; **f)** immunization scheme.

Three unifying tools are utilized:

- **Single clinical history:** used by the general pediatrician and specialists to record check-ups, intercurrent conditions, tests requested and results, work plans, etc.
- **Baby journal**: used to record the child's unique, unrepeatable history; growth and development data; medical indications; intercurrent conditions; medical emergencies; questions and doubts discussed with the physician; memories; words, photos, and drawings of family members, etc. These are rather like *ad hoc* medical records (19).
- **Parents Notice Board**: information is posted on activities and places of interest (grassroots associations; neighborhood clubs; cultural centers: sports, leisure and recreational activities; workshops; courses; free shows for children; trips; visits to museums, etc.).

3. Parents meetings: These are led by the various professionals involved. They are split into two groups in order to enhance the dynamics and focus on the needs of each stage: babies (1 to 12 months) and toddlers (1 to 3 years). Meetings are monthly and last one and a half hours. The topics covered are: language (expression and comprehension; acquisition, etc.); nutrition (right food for every stage; patterns; budget cooking); genetics (popular beliefs; fantasies; guilt/blame; degrees of DS, etc.); pediatric dentistry (a healthy smile for a better quality of life); pediatric clinic (baby care, immunization, breastfeeding, child-rearing guidance, etc.); toy-making workshop (using recycled and cheap materials and aimed at every stage in the child's development and changing interests); sibling meetings (worries about DS, experiences, feelings, identifying with other children in similar situations); social services, etc.

4. Early stimulation: Therapists assess each baby's level of development in various areas (psychological, motor, cognitive, language and communication) and the child is given games, actions and postural exercises that entail new challenges.

The sessions are held weekly and last for 40 minutes.

5. Socialization Group: The Socialization Group is part of Early Educational Psychology (EEP) and is aimed at children aged from 32 months to 4 years who have finished ES and are going through a period of separation/individualization from their parents. Its goal is to integrate them successfully in a kindergarten, creating a link between the Atiendo Group and the school institution. The areas covered at this stage are: psychomotor (locomotion, ability to explore space); cognitive (sensory-motor skills, access to the symbolic function); creation of a sense of self (differentiating oneself from mother and others); language (use of 'No', starting to use pronouns); habits (some independence).

Meetings are held weekly and last one hour.

6. Social Services: All parents of patients on the program are interviewed by the Social Services where they are told how to get a transport pass and disability allowance.

4. Results

Between 1/1/1996 and 6/30/2002 of a total of 41,747 live births, 98 newborns were diagnosed with DS (2.34/1,000) (Table I).

Sex: 56/107 boys; 51/107 girls.

Birth weight (BW) average: 2,939 grs.± 0.505

Gestational age (GE) average: 38.4 wks. ± 2.0

Breech presentation: 11/107 **C-sections**: 30/107

Average age of mother: 33.5 years. ± 7.5. **Range:** 15-44 years

Home remote (> 100 km.): 5

Referrals to surgery (< 48 hrs.) for heart or digestive disease: 5

Deaths during neonatal period*: 2* **Deaths** < 12 months: 4

Deaths aged between 1 and 2 years: 4

Conditions diagnosed on birth or later:

– Heart disease (by Eco-Doppler): 47/107 children (43.9%) were diagnosed, the most common being ventricular septal defect (VSD) and atrial septal defect (ASD), associated or alone (**Figure 1**).

– Digestive impairments: 12/107 (11.2%) were diagnosed; the most frequent: duodenal atresia **(Figure 2).**

– Urinary impairments: On birth, 1 child with hydronephrosis.

– Ophthalmologic disease: On birth 3 children were diagnosed with cataracts and 1 with megalocornea. During the first year: 5 children with nystagmus. From 2 to 3 years: 5 with strabismus and 7 with myopia.

– Hearing loss (assessed on the basis of brain stem potentials and behavior). Conductive hearing loss: 5. Sensorineural hearing loss: 2.

– Hypothyroidism: Detected in 2 children, one with congenital hypothyroidism and another at age 2 (1.86%).

– Neurological dysfunctions: West syndrome, hypsarrhythmia or disorganized EEG: 7/107 (6.54%).

- **Karyotype:** A karyotype was done on 80% of the children. The most common form is the free trisomy 21 (47, XX or XY, + 21) and in 5 children (4.6%) a translocation was found.
- **Growth:** Average weight, height and cephalic perimeter at 1, 2 and 3 years was:

Five (5) girls and one (1) boy were above percentile (P) 95. On average, they were between P50 and P75 on the growth charts for children with DS **(Figures 3, 4, 5, 6).**

- **Social and family issues:** Of all 107 children, in 64.48% (69 children) the parents stayed together, in two cases (1.85%) the mothers became widows during pregnancy, and in the remaining 34.5% (37 children) the parents split up after the birth or during the baby's first months.

With regard to disability allowance, 86% had not yet received it (still being processed), 12.14% were receiving it and 1.85% received social assistance.

5. Discussion

Secondary prevention (early screening and intervention in children with a defined condition) prevents deterioration and more severe disabilities in the future. The higher rate of children with DS (approximately 2.34‰) as compared to the literature could be attributable to the HMIRS's status as a referral centre for high-risk pregnancies.

The rate of heart disease (43.9%) was similar to that found in the literature (30 to 50%) (8, 9). The most common heart diseases were VSD and ASD, alone or in association, while published references name atrioventricular septal defect as the most frequent heart disease.

The most common digestive condition found was duodenal atresia, which matches findings by other authors (16).

Growth between birth and 3 years compared well to the growth curves for American children with DS (3). (No growth charts are currently available for Argentinean or Latin American children with DS.)

Seven children presented infantile spasms or West syndrome, a relatively common condition in DS. Some authors use the term hypsarrhythmia generically, because of the type of electroencephalogram obtained (14). The reason for its higher incidence in this population is not known.

One significant difference is the low mortality recorded at one year in children with DS in our population (9.3%) compared to Latin American populations, according to the Collaborative Latin American Study of Congenital Deformations (ECLAMC: 20%) (20), which underscores the importance of early detection and treatment of associated illnesses.

Hypothyroidism is a commonly occurring condition in children with DS (21). We detected only 2 cases: 1 congenital and the other acquired.

The fact that 63% of the children attend ES increases the likelihood of their starting school earlier, and in good conditions.

This study highlights the strain that bringing up a child with a disability can put on a relationship: in 34.5% of cases (37 children) the parents split up straight after the birth or during the child's first few months.

Table I.
Newborns with DS diagnosed between 1-Jan-1996 and 30-Jun-2002

Year	Total live births	Children with DS
1996	7,188	16
1997	6,809	17
1998	5,950	10
1999	6,078	11
2000	6,341	18
2001	6,004	17
1er semestre 2002	3,377	9
TOTAL NB	**41,747**	**98**

Figure 1. Heart disease (by Eco-Doppler).

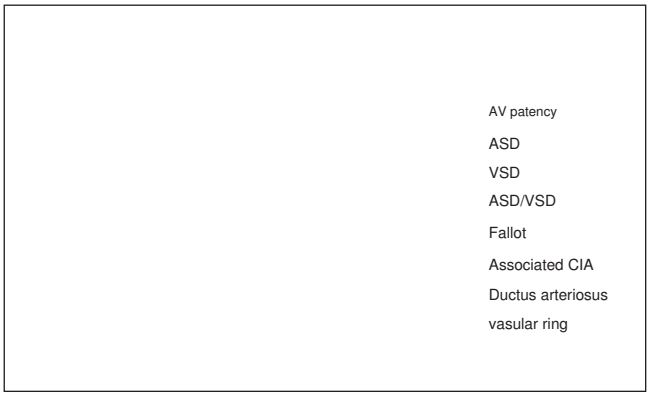


Figure 2. GI tract malformations.

Table II.
Average weight, length/height and head circumference at birth and 1-2-3 yrs

Boys:	Birth	1 year	2 years	3 years
Weight (g) (X) DS	2,885±0.68	8,635±1.02	11,082±1.22	12,901±1.40
Lenght (cm) (X) DS	48,04±2.6	70.77±2.84	80.17±2.83	88.05±3.64
CP (cm) (X) DS	33,88±1.5	44.51±1.33	46.91±1.43	48.50±1.72

Girls:	Birth	1 year	2 years	3 years
Weight (g) (X) DS	2,939±0.5	8,329±1.12	10,635±1.05	12,616±1.39
Lenght (cm) (X) DS	47.78±1.96	70.30±2.55	79.42±3.04	86.66±3.35
CP (cm) (X) DS	33.77±1.17	43.77±1.54	46.05±1.44	47.49±1.26

SD: standard deviation

At the time of the study, 86% of the children were still not receiving an allowance, a figure that matches nationwide statistics according to an ENDI-INDEC (National Disability Survey) poll which reveals that while in 20.6% of households in Argentina there is at least one individual with a disability, 81% of these households get no disability allowance.

Although benefits and/or grants are not easy to claim, networks for solidarity, friendship, cooperation and mutual help are being set up: "whenever one door closes another opens".

The program drop-out rate is 7.4% (8/107) for children under the age of 2, which is lower than other HMIRS follow-up schemes (12). From 2 to 3 years this rises to 14% (15/107), particularly in children who were not born at the HMIRS, probably due to their starting kindergarten. Attendance is voluntary, with travel expenses paid through the Volunteer Service and a ration of milk, iron and vitamins donated by the HMIRS.

All illnesses and conditions have unique characteristics that set them apart from other illnesses or disorders. If these young patients are to achieve maximum growth and development, intense concentration and interdisciplinary coordination is needed in monitoring and following them up. These children have complex issues requiring care over a whole range of areas so they can stay together. Any other approach would only enable them to make progress on issues shared by all.

The Atiendo Group may seem to be 'different' from other groups – a kind of 'web' that entraps the person with a disability in a dead end and cuts them off from the rest of the community. In fact, the aim of this activity, which some may regard as overly protective, is transitional, and its function is to provide the tools and security needed so that the children and their families can make progress both in personal independence and integration into society.

Figure 3. Weight curve for boys with DS at HMIRS (1996/2002).

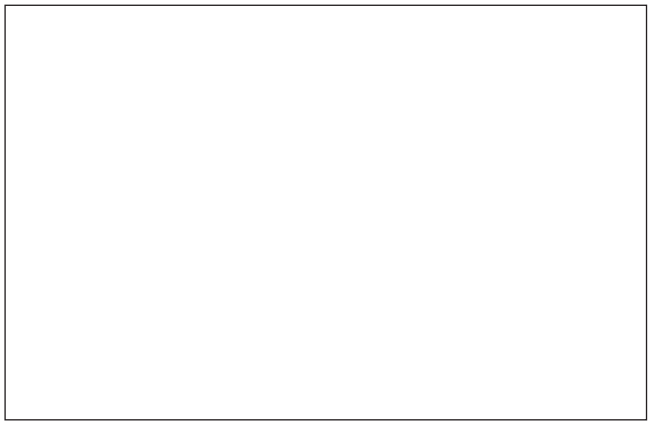


Figure 4. Height curve for boys with DS at HMIRS (1996/2002).

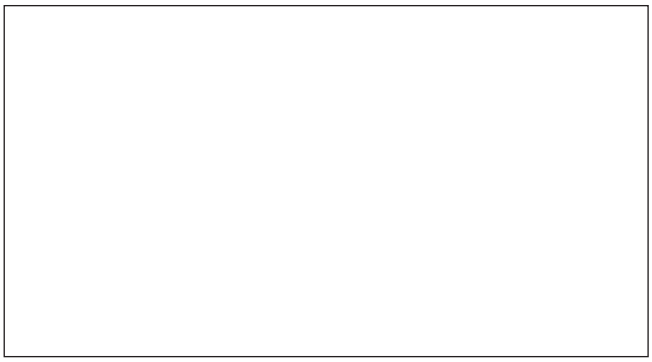


Figure 5. Weight curve for girls with DS (1996-2002).

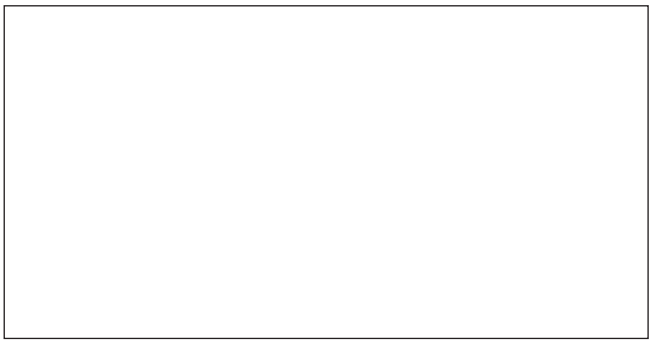
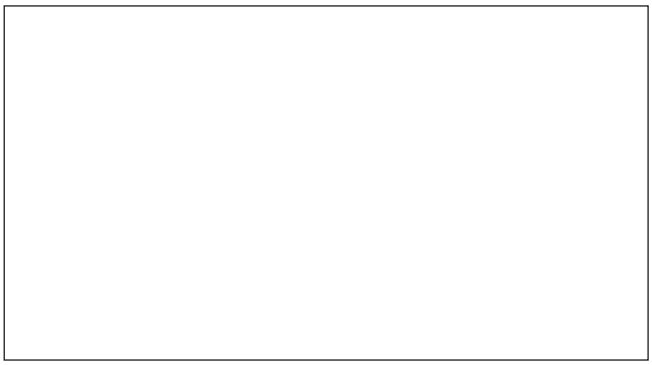


Figure 6. Height curve for girls with DS (1996-2002).



6. Conclusions

A follow-up program such as this aids earlier detection of conditions and increases attendance at follow-up and ES sessions, making for better family and social integration. The various activities that the Atiendo Group encompasses give children with DS a chance to reach their maximum development potential, increase survival, and enhance their quality of life, social and family integration, as the targets are met. Moreover, the ethical component of a task that entails problems linked to these values must not be overlooked. If these are properly applied and respected they contribute to a successful professional/ patient/family relationship. Taking a holistic approach to a child with DS or any other disability means treating them first and foremost as a person. The interdisciplinary team enriches everyday activities and patient care at large, as well as every member of the team itself.

Our final word: We have chosen this path because we wish to walk alongside our patients. There are needs and shortcomings that we pledge to resolve because we have a commitment to life and seek to face up to its challenges.

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