ORIGINAL ARTICLE

Down Syndrome health care protocol - IMREA/HCFMUSP

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ABSTRACT

The Down syndrome (DS) or chromosome 21 trisomy is the most common chromosomopathy in human beings, it occurs regardless of gender, ethnicity, or social class. In Brazil, there is approximately one child born with DS for every 700 births. It is known that people with Down syndrome well cared-for and stimulated have potential for full social inclusion. This protocol was prepared by the Down Syndrome Personal Health Care multiprofessional team at the IMREA/HCFMUSP. **Objective:** Is to offer orientation in the health care of a person with Down Syndrome, in the different levels of attention to health, throughout his/her life. **Method:** The preparation of the total care protocol for the health of a person with Down syndrome was based on searches in the PubMed and SciELO systems and on the Cochrane Database of Systematic Reviews using the keywords: Down syndrome and Syndrome of Down, Trisomy 21, "Trisomía del Cromosoma 21", Chromosome 21 trisomy, Growth, "Desarollo", and "Crescimento". **Results:** The articles reviewed were published from 1972 to 2011 and limited to the languages: English, Spanish, and Portuguese. Records previous to 1972 were also included for being considered historical. **Conclusion:** The data was analyzed by a group of specialists that discussed the results and prepared this protocol.

Keywords: down syndrome, patient care team, rehabilitation centers

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INTRODUCTION

The first clinical description was made in 1866 by John Langdon Down, who worked at the John Hopkins Hospital in London in a care facility for people with intellectual deficiency; in his studies he classified these patients according to the phenotype.[1] He called those individuals with short stature, straight hair, upward-slanting palpebral fissures, flat nasal bridge, and with slight to moderate intellectual deficit "mongoloids". Because it was the first complete description of a category of patients, this set of signs and symptoms was named Down Syndrome, in recognition of Langdon Down. Unfortunately, the term "mongolism", which is now in disuse, was also assimilated as a currently used term; due to its pejorative connotation it is considered archaic and prejudicial and must not be used. In view of this discordance, the term "mongolism" was stricken from the Lancet Magazina in 1964, from the World Health Organization publications in 1965, and from the Index Medicus in 1975. Waardenburg (1932) suggested that the occurrence of Down Syndrome (DS) was caused by a chromosomal aberration; two years later Adrian Bleyer proposed it to be a trisomy. This was confirmed two decades later, in 1959, by Lejeune and collaborators, who demonstrated the presence of the extra chromosome 21 in DS.[2] In 1960, the first cases of translocation were described by Polani, and in 1961, the first case of mosaicism.[3]

The life expectancy of people with DS has increased considerably from the second half of the 20th century due to progress in the health area - especially cardiac surgery. The increase in survival and in understanding the potential of people with Down Syndrome has led to the development of different educational programs, towards schooling, a professional future, autonomy, and quality of life. More and more society is becoming aware of how important it is to value human diversity and of how fundamental it is to offer equality of opportunity so that people with deficiencies can exercise their right to live in the community.

Despite the experience accumulated in recent years, it is not possible to foresee the degree of autonomy a child with DS will have in its adult life. The potential to be developed is always a frontier to be crossed daily. Every investment in health, in educational programs, and in social inclusion results in better quality of life and autonomy.

Concept

The term "syndrome" refers to a set of signs and symptoms and Down designates the name of the physician and researcher who first described their association. Down Syndrome means, therefore, a set of signs and symptoms described by John Langdon Down in 1866, whose etiology was elucidated by Jerome Lejeune in 1959, as trisomy of the chromosome 21.[1,2] It is important to point out that the Down syndrome is not a disease or pathology, but a genetically determined human condition, a way of being in the world that demonstrates human diversity.

Clinical Diagnosis

The clinical diagnosis of DS is based on the identification of a set of characteristics.[3,4,5] Down Syndrome has its clinical appearance explained by an imbalance of chromosomal constitution, the trisomy of chromosome 21, by simple trisomy, translocation, or mosaicism. Despite the existence of three possibilities of genotype, DS presents a common phenotype with a range of intensities. We understand genotype to be the chromosomal constitution of the individual, and by phenotype, characteristics observable in the organism that result from the interaction of genic expression and environmental factors.

The DS phenotype is characterized mainly by: upward slanting palpebral fissures, epicanthus, unibrow (union of the eyebrows), flat nasal bridge, acromicria (hypoplasia of the middle third of the face), tongue protrusion, ogival palate, small ears with lower positioning, thin hair, short fifth finger with clinodactyly, brachydactyly, single transverse palmar crease, separation between the 1st and 2nd toes, flat feet, hypotonia, ligament laxity, and umbilical hernia.[3,4,5] These signs may not be present in their totality, but they are noticeable in various associations and expressions (Table 1).

As for the psychomotor and pondero-statural development, the literature affirms that 100% of people with DS show some deficiency in these aspects. The intellectual deficit of people with DS ranges from mild (IQ 50-70) to moderate (IQ 35-50), and is rarely severe (IQ 20-35).[5]

According to publications in the Pediatric Database[5] and by the American Academy of Pediatrics Committee on Genetics,[6] there is a set of alterations associated with DS that demands special attention and needs specific exams for its identification, they are: alterations in hearing, sight, orthodontics, endocrinology, in the locomotor system, in the digestive system, neurological, and hematological systems, in addition to congenital cardiopathies (Table 2).[6] Domestic studies also revealed high prevalence of celiac disease (5.6%) in children with DS, confirmed by molecular investigation (Alleles DQ2 or DQ8) and/or research of antibodies (anti-endomysial, anti-gliadin, and anti-transglutaminase), and/or biopsy.[6-10]

Laboratory Diagnosis

The laboratorial diagnosis of Down Syndrome is done through the genetic analysis called karyotype. The Karyogram or karyotype is the representation of the set of chromosomes present in the nucleus of the cells of an individual. In the human being, there are 23 pairs of chromosomes, that is, 46 chromosomes, with 22 pairs of chromosomes called autosomes and one pair of sexual chromosomes, represented by XX in females and XY in males. In the karyotype the chromosomes are ordered by descending order of size. The Down Syndrome is characterized by the presence of an extra 21st chromosome that can present itself as simple trisomy, translocation, or mosaic.[3,11,12] The karyotype is not obligatory for the diagnosis, but it is fundamental for genetic counseling.

Simple trisomy (95% of the DS cases) occurs casually and is characterized by the presence of an extra free 21st chromosome.

In translocation (3 to 4% of the DS cases) the genetic material of the 21st chromosome results from an unbalanced translocation and it is connected to the acrosome of another chromosome, most frequently the 14th chromosome. Its occurrence is associated with the presence of familial translocation- that is, one of the child's progenitors had a balanced translocation, which resulted in a gamete

Table 1. Clinical diagnosis of DS based on the following characteristics

Segmental exam		Signs and symptoms
Head	Eyes	Epicanthus
		Upward slanting palpebral fissures
		Unibrow
		Brushfield spots
	Nose	Flat nasal bridge
		High palate
	Mouth	Hypodontia
		Tongue protrusion
	Form	Brachycephaly
	Hair	Thin and placed low
	Ears	Small with delicate lobe
		Low positioning
Neck	Connective tissues	Excess adipose tissue on the nape of the neck
		Excessive skin on the neck
Thorax	Heart	Cardiopathy
	Nipples	Mammary hypertelorism
Abdomen	Abdominal wall	Diastasis of the rectus abdominis muscle
	Umbilical scar	Umbilical hernia
Locomotor System	Upper	Single palmar crease
		Clinodactyly of the 5th finger
	Lower	Distance between the hallux and the 2nd toe
	Tone	Hypotonia
		Ligament laxity
Development	Intellectual deficit	
	Psychomotor deficit	
	Pondero-statural deficit	

Adapted from the American Academy of Pediatrics Committee on Genetics (2011)[5]

(sperm or egg) with an extra 21st chromosome. In these cases there is a greater chance of familial recurrence of the DS.

Mosaic (1 to 2% of the DS cases) occurs by chance and is characterized by the presence of two cellular lineages, one normal with 46 chromosomes and another trisomic with 47 chromosomes.

In the International Classification of Diseases (CID-10) the Down Syndrome receives the code Q-90, because it is classified in chapter Q00 - Q99 of malformations, deformities, and chromosomal anomalies. In this chapter chromosomal anomalies are found in the Q90 - Q99 group and the Down Syndrome is found in the Q90 category.[13] In the Q90 category there are the following sub-groups:

Q 90.0 - Down Syndrome, trisomy 21, for meiotic non disjunction.

Q 90.1 - Down Syndrome, trisomy 21, mosaicism for meiotic nondisjunction.

Q 90. 2 - Down Syndrome, trisomy 21, translocation.

Q 90. 9 - Down Syndrome, not specified.

Prenatal Diagnosis

The pre-natal diagnosis of DS is possible from the first trimester of gestation. The pre-natal evaluation in the first semester includes: morphological ultrasound, nuchal translucency evaluation, evaluation of the proper nose bones, dosage of Human Chorionic Gonadotropin (β-hCG) and of Plasma Protein A (PAPP-A) in the mother's blood. The nuchal translucency is an ultrasonic hypoechogenic image of liquid accumulation in the back of the neck, which occurs frequently between the 10th and 14th weeks of gestation, possibly by alteration of the cervical lymphatic drainage or hemodynamic disturbances.[14,15,16] In a multicentric study involving 100,000 pregnant women, it was observed that 72% of the fetuses with chromosome trisomy 21 had the measurement of nuchal translucency above 95%, which led to the conclusion that this exam has strong predictive value for chromosomal anomalies.[17]

Fetal anthropometric studies demonstrated that in 60 to 70% of the fetuses with chromosome trisomy 21, the nasal bone was not visible in ultra-sound between 11 and 14 weeks of gestation. High levels of Human Chorionic Gonadotropin (β-hCG) and low levels of Plasma Protein A (PAPP-A) in the maternal serum, added to the altered measurement of nuchal translucency detect 86% of gestations with DS fetuses.

In the second semester, between the 15th and the 20th week of gestation, the triple test with dosage of alpha-fetoprotein (AFP), Human Chorionic Gonadotropin (β- hCG), and non-conjugated estradiol (uE3) in the maternal serum is performed. When the dosage of inhibin is included it is called the Quadruple test. High levels of b-hCG and low levels of AFP and uE3 suggest DS in the fetus, and the values must be related to the time of gestation. The inhibins are glycoprotein hormones synthetized by the placenta and have stable concentration during the second semester of pregnancy, but are increased in the DS. The pre-natal diagnosis scheme detects DS in 95% of the cases, with 5% of false-positives (Table 3). The data obtained leads to confirmatory exams such as chorionic villus biopsy, amniocentesis, and cordiocentesis.[14-17]

The pre-natal diagnosis can be confirmed by analyzing the chromosomal constitution of the placenta, done through chorionic villus biopsy, during the 10th and 12th weeks of gestation. The chromosomal constitution of the fetus is analyzed in cells dispersed in the amniotic liquid, through a procedure called amniocentesis or by the analysis of the fetus' blood contained in the umbilical cord, which is performed in the second semester of gestation, after the 15th week and the 18th week of gestation, respectively. These exams are invasive and offer risk of miscarriage, therefore they have restricted recommendation.[18]

Once the diagnosis is confirmed, genetic and pediatric counseling must be initiated still in gestation, with guidance about the prognosis, global stimulation programs, and future treatments, in addition to guidance on social support networks such as associations, parents' groups, and community resources.

Table 2. Pathologies associated with DS and their prevalence

Systems	Pathology	Prevalence
Visual system	Cataract	15%
	Lacrimal duct stenosis	2%
	Refractive error	50%
Auditory system	Auditory loss	75%
	Chronic otitis	50-70%
Cardiovascular System	ASD	40-50%
	VSD	
	AVSD	
Digestive System	Esophageal atresia	12%
	Duodenal stenosis/atresia	12%
	Hirschsprung disease	1%
	Celiac disease	5%
Nervous System	West syndrome	1-13%
	Autism	1%
Endocrine System	Hypothyroidism	4-18%
Locomotor System	Cervical subluxation w/o lesion	15%
	Cervical subluxation with medullary lesion	1-2%
	Hip luxation	6%
	Appendicular instability	Nearly 100%
Hematologic System	Leukemia	1%
	Anemia	3%

Adapted from Pediatric Database (1994)[5] and from the American Academy of Pediatrics Committee on Genetics of the (2011)[6]

Table 3. Predictive data of Down Syndrome in pre-natal period

Exams	Results
Nuchal translucency	≥ 2.5 and 3 mm
	≥ percentile 95
Nose bones	Absent or hypoplasic
Triple test	β-hCG ≥ 2.0 MoM
	AFP ≤ 0.5 MoM
	uE3 ≤ 2.5 MoM
Plasma Protein A	Diminished
Inhibin	Elevated
Chromosomal analysis of the chorionic villus	Chromosome 21 trisomy
Amniocentesis	Chromosome 21 trisomy

Moment of Breaking the News

The birth of a child with DS is, in general, marked by difficulties for the parents, siblings, and family, especially amplified by the lack of proper information and lack of preparation of health professionals.[6,19,20,21]

The uncertainties and insecurities are many, as much for health and immediate development potential as for possibilities of future autonomy and quality of life. Such feelings blend with the difficulty in accepting a child who was born different from their hopes.[22,23]

The support of qualified professionals is fundamental for the family adjustment to the new situation, which favors the possibilities of treatment leading to the physical, mental, and emotional health of the child.[24]

In this sense, the moment of the news of the birth of a child with DS impacts the acceptance of the family and its willingness for and adherence to treatment. It is expected of the professional to break this news in a human and ethical way, which guarantees the welcoming and informing of the family.

The following directives are recommended for broaching the DS diagnosis to the family:

- Broaching the diagnosis must be done when the presence of the characteristic phenotype is confirmed by more than one member of the team or by confirmation of karyotype.
- The news must be given in the first 24 hours in case of newborn and preferably outside the birth room.
- The pediatrician is the person responsible for conveying the diagnosis, and may be helped, in this moment, by the obstetrician who did the pre-natal monitoring. This responsibility must not be "passed along" to another member of the multiprofessional team.
- Communicating with the mother is preferably done in the presence of the father, or, in his absence, of another member of the family who represents a significant relationship.
- The location must be reserved and protected from interruptions.
- The pediatrician must have time available to convey the diagnosis, and give the prognosis, treatment, and other DS characteristics, in addition to listening and allowing the family to exhaust its questions and express its feelings.
- Starting with the first contact, one should congratulate the parents and call the baby and the parents by their respective names.
- During the physical exam show the parents the phenotypical characteristics of DS that led to the clinical diagnostic.
- It is important to highlight that the word "syndrome" means a set of symptoms and "Down" is the name of the man who described it for the first time. Thus, dissolve the family anguishes a little while facing the stigma of a syndromic child.
- Avoid the word "sufferer", for DS is not a heavy burden someone

suffers from, but a different way of being in life, that is, a life condition.

- Discussing the etiology with the parents is important so they can resolve some questions and feelings of guilt. The etiology must be approached showing a karyotype and explaining its constitution, which in the case of DS contains an extra chromosome 21, which is responsible for the clinical DS characteristics. Due to the presence of this extra chromosome 21 in all the cells, until now there is no cure for DS, but it is important to point out that there is treatment and that it is decisive for more autonomy and quality of life in the future.
- Emphasize that the caring for the baby be shared between the family and the multiprofessional team, and that the family will not be alone and without support in this process.
- The pediatrician must finish this first conversation with the family assuring his availability to them.

The moment of breaking the news of the birth of a child with DS is a health education process, in which the physician teaches and guides the family. In this first contact with the family it is not recommended that the pediatrician report all the pathologies that a person with DS may present during his or her lifetime, in the same way that it is not done in the birth of a child without DS. The information must be offered according to the demands of the family; it must be realistic and focused on the potentialities of a person with DS.

Genetic Counseling

In the understanding of the American Society of Human Genetics,[24] genetic counseling is a communication process that deals with human problems associated with the occurrence, or risk of occurrence, of a genetic alteration in the family, involving the participation of one or more trained people to help the individual or his/her family to understand the medical facts, including the diagnosis, the probable course of the condition, and the available courses of action, the way in which heredity contributes to the condition and the risk of recurrence for specific relatives, and choose the course of action that seems most appropriate due to the risk, family goals, ethical and religious standards, acting in accordance with this decision and adjusting in the best way possible to the situation imposed by the occurrence of this disturbance in the family, as well as to the prospect of its recurrence.

Genetic counseling concerning DS must initially aid the family in the understanding of its etiology and of the clinical and laboratory diagnosis. Showing the karyotype to the family and explaining the image and report is fundamental to understand the etiology of DS, and also important to minimize the guilt feelings that the family may experience. It is important to note that Down Syndrome occurs in most cases (95%) due to a genetic imbalance during the gametogenesis- that is, in the formation of the sperm and of the egg. Instead of 23 chromosomes, one of the gametes that generated the child with DS brought 24 chromosomes - that is, it brought in with its set of chromosomes one extra chromosome 21. This situation results in simple or free trisomy; its chances of occurrence or recurrence are less than 1%. However, it is important to remember that the risk of recurrence increases with the mother's age, and can reach 4.5%.[6,10,11,25]

The cases of mosaicism, which correspond to 1 to 2% of the DS cases, carry normal and trisomic cells in their genetic constitution, which are also of chance occurrence, but occur after the fertilization, during the first cell divisions of the embryo. For cases of mosaicism, the chance of recurrence is also less than 1%.[6,10]

Familial recurrence is higher in the cases in which there is chromosomal translocation, approximately 3 to 4% of the DS cases. In these cases the genetic material of the chromosome 21 is connected to another chromosome, most often the chromosome 14. Facing the diagnosis of chromosomal translocation, a genetic analysis of the progenitors is recommended, due to the possibility of one of them having a balanced translocation- that is, despite not having an extra chromosome 21, his/her chromosome 21 is connected to another chromosome, in an abnormal position. If this is the case, the progenitor does not have DS, but can generate gametes with an extra chromosome 21. In these cases the chance of recurrence is 12 to 16% if the carrier of the translocation is the mother, and 3 to 5% if it is the father. If no translocation is found in the progenitors, then the translocation happened only in the gametes, which brings the chance of recurrence to 2 or 3%.[25]

Another question to be broached in genetic counseling is the association of DS with the advanced age of the mother. Many studies have demonstrated the increase in the incidence of DS in gestations in which the mothers are older than 35 years; the incidence of trisomy in gestations of mothers younger than 25 years is 2% and 35% in mothers older than 40 years. This is due to the aging of the gametes, its chromosomes and spindle apparatus, in addition to the diminution of uterine protection factors that would recognize an abnormally constituted zygote, making it difficult to implant it. Some studies describe an increase in the incidence of DS in gestations where the father was older than 55 years.[27]

Lastly, genetic counseling must give guidance on the therapeutic possibilities. In this consultation, some professionals give the parents a list of the pathologies associated with DS, even those associated with maturity, such as early aging and Alzheimer risks. This information has the potential to generate anguish and uncertainty about the future of the family. Counseling must be to attend to immediate health questions according to the age of the person attended, leaving the other questions for subsequent consultations. The focus of the orientations beyond the health diagnosis questions (associated pathologies) on the child from zero to three years old should be global stimulation and the acquisition of psychomotor skills, between four and five years old, questions of socialization and behavior, and between six and 12 years old, questions of schooling. After 13 years of age the counseling should focus on autonomy, sexuality, and vocational orientation. For youths and adults the questions of autonomy and employability, as well as planning the future should be approached.[6]

Health Care

Health care at the Outpatient Clinic for Personal Care of People with Down

Syndrome at the Lucy Montoro Network is guided by public policies from the Ministry of Health such as Humanization National Policy,[28] Children's Health Programs[29] and of the Adolescent,[30] Women's,[31] Men's,[32] and Elderly's[33] Health, Mental Health,[34] and in the World Report on Deficiency.[35]

The presupposed theories of an extended, holistic, shared-care clinic are used, leading to the humanization, autonomy, and support of the subjects in health practices.[36,37] In addition to integrating the bio-psycho-social focuses, the extended clinic articulates the service network of the Unified Health System and the resources of the community. The practice of the extended clinic is transdisciplinary and considers the complexity of the subject's life in which the process of disease, care, rehabilitation, prevention, and the promotion of health evolve. It demands reorganizing the service, reviewing the practices, and preparing protocols. Health work, in the ambit of the extended clinic, demands that its professionals respect and share multiple knowledge, communicate, be flexible, and be responsible for the patient.[28]

Holism as a theory of the extended clinic and of the personal care for people with DS may be understood as quality of the care, as a way of organizing the practice, and as a government answer to the community health problems. Admittedly holism is an aspect of good practice in health care, and a value to be preserved, since it does not reduce the individual to merely biology, but it expands the view of those who attend to the psycho-social -emotional dimensions of the patient, including also the aspects of prevention, health promotion, and education.[38]

Finally, shared care relates to the multiprofessional teamwork that builds the diagnosis, the therapeutic project, defines therapeutic goals, and jointly re -evaluates and monitors the therapeutic process. However, shared care can also be understood as an integration of the different densities of technologies of attention to health in the Health System, as well as their integration with the community resources. Sharing care is also the co-responsibility of the care process among professionals, the subject under care, and his/her family.

Personal care for the person with DS follows the guidelines below:

1. Expanded understanding of the health and disease processes;
2. Shared skill building by the multiprofessional team of a Situational Diagnosis, which involves, in addition to the clinical and general health diagnoses, the psychomotor evaluation, language development, social-emotional behavior, and a survey of needs and potentialities of the child, aside from the available therapeutic resources;
3. Shared skill building of Individual Care Plan that considers bio-psycho-social aspects of the patient and the family and community resources;
4. Shared definition of the Therapeutic Goals;
5. Commitment by professionals, family, and the individual to the therapeutic goals.

The health care of the person with DS at the Lucy Montoro Network, as an example of an extended clinic that deals in health under the logic of holism and of shared care, is supported by a multiprofessional team with the following professionals: physician, dentist, nurse, psychologist, nutritionist, social worker, physical therapist, occupational therapist, speech therapist, physical trainer, and an educator. The other medical specialties are supplied as needed by the physicians at the Clinical Hospital of the Medical School at the University of São Paulo. Attention to health in DS demands a holistic view in caring, and constant conversation with different specialists. In this sense, the actions of the physician must be holistic, avoiding thus the conspirary of anonymity, that is, a situation where the person with DS is seen by different specialists who do not talk among themselves and many times do not meet the demands of the patient, because they think that this is already being done by another specialist.

Health care in DS in our service is segmented into models according to life stages. In each stage, the attendance seeks to maintain health to reach better development of the potentialities of the person with DS, aiming at quality of life and social and economical insertion.

Believing that the health of a person with DS is directly related to his/her living habits, the work of health professionals is directed towards the promoting healthy life styles in the nuclear family. For that we utilize health education strategies with the family, relying on its support and autonomy to share the care for the person with DS, also promoting health in this family nucleus.

The following health care models are offered:

The *Global Stimulation Model,* which receives children from zero to three years old, and has its focus on acquiring motor skills. The child with DS needs stimulation as soon as he or she is diagnosed. From this standpoint, despite the frequent use of the term "precocious stimulation", it has not been adopted in our service for we consider that intervention in DS cases is necessary and not precocious, that is, it is expected to start right after the diagnosis. Therefore, in this case, we recommend designating this attendance as "global stimulation" or simply "stimulation".

The *Child Development Model* is for children from four to eleven years of age, focusing on the acquisition of social abilities, autonomy for daily life activities, schooling, and improvement of balance and mobility.

The *Adolescent Model* is directed to youths aged from 12 to 18 years and seeks to develop autonomy, self-care, and independence for instrumental daily life activities.

The *Adult Model* receives people older than 19 and focuses on autonomy, socialization, employability, and planning the future in relation to financial support and care along the course of life.

For each one of these models the attendance flow calls for an initial evaluation of each one of the specialties, followed by a discussion of the case by the team that jointly prepares the Situational Diagnosis and the Individual Care Plan.

Some care and stimulation services for people with DS offer assistance by an essential therapy team consisting of: a physical therapist, a speech therapist, and an occupational therapist. Our experience and work philosophy has made our therapy team expand to include other professionals: psychologist, physical educator, nutritionist, and educator. In addition there is a support team: physician, nurse, dentist, and social worker. The therapy team examines the patient and their family weekly, and the support team monthly, bimonthly or every six months.

Health care from zero to three years old

Health care for the child with DS must be focused on support and information for the family and on the diagnosis of associated pathologies. After this initial phase the Individual Care Plan includes global stimulation, immunization, stimulus to breast feeding, and maintenance of health with periodic monitoring.

After informing them of the birth of a child diagnosed with DS, the pediatrician must guide the family and request the necessary supplementary exams: karyotype, echocardiogram, complete blood count, TSH (Thyroid Stimulating Hormone), and thyroid hormones (T3 and T4).[39]

Karyotype is the exam requested for laboratory diagnosis of DS, and it must be requested on the first day of life, or if it had not been done, at any time afterwards.

The echocardiogram is requested because 50% of children present cardiopathies, the most common being: Atrial Septal Defect (ASD), Ventricular Septal Defect (VSD), and complete Atrioventricular Septal Defect (CAVSD). In case the first exam is normal, it is not necessary to repeat it, but maintain clinical monitoring. Children with cardiopathy must be monitored by a pediatric cardiologist.

A complete blood count is requested to eliminate hematological disturbances such as leukemoid reactions, polycythemia, leukemia, and Transitional Myeloproliferative Disorder, which afflict 10% of newborns. The complete blood count must be done annually over the course of life of the person with DS.[5]

The thyroid function (TSH and free T4) must be evaluated at birth, at six months old, at 12 months old, and every year thereafter. There is a 1% risk of congenital hypothyroidism, and a 14% risk of hypothyroidism in the course of life.[39]

In this initial monitoring phase, pathologies associated with the digestive system must be eliminated, such as: esophageal atresia, duodenal membrane, and Hirschsprung disease. Also common are: constipation, gastroesophageal reflux, and cholelithiasis.[40] A case of constipation at any age the following must be evaluated: ingestion of liquids, hypotonia, hypothyroidis, gastrointestinal malformations, and Hirschsprung disease.

Evaluations of auditory and visual acuity are necessary at six months old and at 12 months old, and then annually in order to prevent refractive errors, congenital cataract, nystagmus, and lacrimal duct stenosis.

As for auditory loss, special attention must be given to Serous Otitis Media, which afflicts 50 to 70% of children with DS and may potentially lead to auditory loss, with repercussion in the acquisition of language and learning. The evaluation of auditory acuity must be done at birth, at six months old, and then annually.

In this phase from zero to three years old it is important to care for repeated respiratory diseases.

Muscular hypotonia is present in 100% of the children born with DS, tending to decrease with age, however the tonus is an individual characteristic and presents variations from one child to another. The presence of hypotonia affects the development of the child, delaying the acquisition of motor competences: supporting its own head, rolling, sitting, dragging, crawling, walking, and running.

In the first infancy and in other phases of life, the family and the patient must be guided on the correct positioning of the neck, avoiding medullary lesion due to the atlanto-axial joint instability. The total flexion and extension movements of the cervical column must be rigorously avoided if performed in somersaults, dives, horseback riding, gymnastics, and during the anesthetic preparation for a surgery. Literature recommends cervical column radiography starting at the age of three when the ossification of this region is complete. This exam must first be done in the neutral position. Values ≥ 4.5 mm of the atlas-axis distance must be considered abnormal and indicative of atlanto-axial subluxation. This situation counter-indicates dynamic cervical column radiography, so a magnetic resonance (MRI) is recommended in these cases. In cases with values ≥ 4.5 mm a dynamic radiography of the column can be done. This type of exam has been discussed due to the number of false negatives and the risk of cervical lesion during the exam. Currently some specialists are not recommending it, keeping radiographic exams for patients who practice sports. Therefore, the current recommendation is to always guide the cervical posture and take radiographies when necessary.[6,8]

In the first phase of life care should be taken towards healthy nutrition, keeping breast-feeding exclusive until six months of age and adding supplementary food up to at least one year. Healthy eating habits must be encouraged early on and whenever possible introduced to the family routine. The monitoring of development follows the Cronk[41] development curves for females and males from zero to eighteen years old. The pondero -statural development curves of Mustachi[42] are also available for children from zero to eight years old.

According to the Brazilian Association of Odontopediatrics, odontological monitoring must start in the first year of life, before the beginning of teeth, around six months of age. In this first contact, the family receives preventive orientation on breast-feeding, the use of baby bottle and pacifier, dietary habits, and oral hygiene. The recommended frequency of visits to the dentist between 12 and 36 months is every three months to monitor the development of the first teeth.[43]

The use of a pacifier, also called a non-nutritive suction, is recommended for children with DS, since it aids in the development of musculature of the face and of the suction itself. Some studies show that the pacifier has a protective effect against the Sudden Infant Death Syndrome.[44,45] However, according to the Brazilian Society of Pediatric Dentistry, its use must be suspended at approximately 24 months, with the maximum limit of 36 months.[43] The Brazilian Society for Pediatrics alerts about the deleterious effects of the incorrect use of the pacifier, which can trigger a decrease in the maternal milk, perioral infections, and alterations in the dental arch.[46]

The Down Syndrome Personal Care Outpatient Clinic totally follows the vaccination calendar from the National Immunization Program (Table 4),[47] including the following vaccines: Varicella at 12 months of age and first dose and Hepatitis A also at 12 months with a booster shot at 18 months. Children up to two years old who were born prematurely, who have congenital cardiopathy, and repeated pneumopathy, whether or not associated with other risk factors, must receive passive immunization with monoclonal antibody against the Respiratory Syncytial Virus, in five consecutive monthly doses.[6]

Table 4. Immunization calendar for zero to three-year-old children with Down Syndrome

Age	Vaccine	Dose
At birth	BCG	Single
	Hepatitis B	1st dose
1 month	Hepatitis B	2nd dose
2 months	Pentavalent (DTP + Hib + HB) (Diphtheria, Tetanus and Pertussis + Haemophilus + Hepatitis B)	1st dose
	Poliomyelitis Oral Vaccine	
	Human Rotavirus Oral Vaccine	
	Pneumococcal Vaccine 10 (conjugate)	
3 months	Meningococcal Vaccine C	1st dose
4 months	Pentavalent (DTP + Hib + HB)	2nd dose
	Poliomyelitis Oral Vaccine	
	Human Rotavirus Oral Vaccine	
	Pneumococcal Vaccine 10 (conjugate)	
5 months	Meningococcal Vaccine C	2nd dose
6 months	Pentavalent (DTP + Hib + HB)	3rd dose
	Poliomyelitis Oral Vaccine	
	Pneumococcal Vaccine 10 (conjugate)	
9 months	Yellow Fever Vaccine (regional recommendation)	Initial Dose
12 months	MMR Vaccine (measles, mumps, rubella)	1st dose
	Pneumococcal Vaccine 10 (conjugate)	Booster shot
	Varicella Vaccine*	1st dose
	Hepatitis A* Vaccine	1st dose
15 months	Trivalent Bacterial Vaccine (Diphtheria, Tetanus, and Pertussis)	1st Booster shot
	Poliomyelitis Oral Vaccine	Booster shot
	Meningococcal Vaccine C	
18 months	Hepatitis A* Vaccine	2nd dose

Based on the National Immunization Program and nomenclature according to the Resolution of Collegiate Board of Directors - RDC nº 61, of August 25, 2008 - National Agency for Sanitary Vigilance - ANVISA. Updated in 2012. * special vaccines that are not part of the calendar

Global stimulation must be initiated as soon as the child's health situation allows. Stimulation in this phase seeks to help in acquiring motor, psychological, and social-emotional skills. There are many models of stimulation composed by individual or group programs, with different health professionals. The therapy team in this phase is composed of a physical therapist, speech therapist, occupational therapist, psychologist, and nutritionist, backed up by the presence of a nurse, a physician, and a social worker.

The health care model for this age bracket is called *Global Stimulation*, with a weekly individual session of 30 minutes, with each one of the therapy team professionals, and monitoring by a physician, nurse, social worker, and dentist monthly or as needed by the patient and their family. Children in the age bracket of zero to three years old, attended by the Global Stimulation model, participate in music workshops and aquatic stimulation activities by the Halliwick method.[48,49] All the activities of the stimulation model are organized to receive the parents together with the children, allowing the stimulation moment to be also one more moment of family interaction and of orientation for parents and care-givers.

Health care from four to 11 years old

The health care of a child with DS from four to 11 years old must be focused on the acquisition and maintenance of a healthy lifestyle (diet, body hygiene, sleeping hygiene, and the practice of exercises), on the development of autonomy and of self-care, on socialization, on the acquisition of social skills, and on schooling.

In this phase, the following exams are necessary each year: complete blood count, TSH (Thyroid Stimulating Hormone), and thyroid hormones (T3 and T4), in addition to visual and auditory acuity.

Since the child is much more mobile in this phase, the prophylaxis of cervical lesion must be very well oriented due to the greater risk of atlanto-axial subluxation in DS, even for asymptomatic children with normal cervical radiographies. Parents and teachers must be warned about the risk of cervical lesions during the practice of sports such as swimming, gymnastics, horseback riding, football, and especially somersaults. These sports must be counter-indicated in the existence of symptoms like cervical pain, weakness, hyperflexion, changes in the intestinal and vesical function. In these cases a cervical radiography in neutral position must be taken first. If this exam shows no alteration, a radiographic study is recommended in flexion and extension of the neck. Radiographies that show alterations in the neutral position are counter-indicative for the dynamic study of the cervical column, and the child must be referred to a specialist immediately.[6,8]

Odontological monitoring must be done every semester, focusing on the possibility of dental eruption alterations, since in these cases hypodontia is common. Dental assistance also focuses on the development of self-care in relation to oral hygiene.[46]

In this age bracket, parents and caregivers should pay attention to sleep apnea symptoms, which include abnormal position in bed, waking up during the night, nasal obstruction, snoring, and sleepiness during the day. The main causes of sleep apnea in children with DS are obesity and adenoidal and palatine amygdala hypertrophy. For these cases a permeability study is recommended of the airways and a polysomnography. The quality and quantity of sleep is important in DS since the presence of sleepiness during the day can generate mood changes, concentration problems, and decreased learning.[50,51]

Another focus of care must be the moisturization and integrity of the skin, which tends to be dry and subject to infections. Daily moisturizing and a neutral soap in the washing of clothes is suggested.

In attending this group there is guidance to prevent physical and sexual abuse, through the development of autonomy and self-care.

The monitoring of development follows Cronk[41] development curves for females and males from zero to eighteen years old. The Mustacchi[42] pondero-statural development curves are also available to DS children from zero to eight years old.

The immunization calendar in this age bracket includes the second booster shot of Trivalent Bacterial (Diphtheria, Tetanus, and Pertussis) and the second dose of the MMR Vaccine (measles, mumps, rubella), and of the Varicella Vaccine (Table 5).[47] The Brazilian Society for Pediatrics suggests also for this age bracket the HPV Vaccine (Human Papilloma Virus) in three doses starting at 9 years old for girls and boys.[46]

The therapy team for the 4 to 11 age bracket consists of a speech therapist, psychologist, occupational therapist, physical educator, and nutritionist, assisted also by an educator who will act as a tutor in schooling matters. The attendance model for this age bracket is called *Child Development*, with a weekly individual 30 minute-session with each one of the therapy team professionals, and monitoring with a physician, nurse, social worker, and dentist, every semester or as demanded by the patient and their family. The children attended in the Child Development model also participate in Dance workshops and Art Experience and in aquatic activities.

Health for the adolescent

Health care for the adolescent with DS must be focused on maintaining a healthy lifestyle (diet, body hygiene, sleep hygiene, and the practice of exercises), on the development of autonomy and self-care, on socialization, on schooling, and on vocational orientation. In this phase they also receive orientation on sexuality and on the prevention of pregnancy and sexually transmitted diseases.

Every year the following exams are necessary: complete blood count, dosage of TSH (Thyroid Stimulating Hormone) and thyroid hormones (T3 and T4), in addition to annual auditory acuity, and bi-annual visual acuity.

In this phase of life, as well as in the previous phases, it is important to maintain the importance of cervical posture orientation and request radiological study of the cervical column in the existence of cervical pain, torticollis, weakness in the upper limbs, dizziness, or intestinal and vesical alterations. In this case a cervical radiography in the neutral position must be taken first. If there are no alterations it is possible to make a radiographic study in flexion and extension of the neck. In the cases with alterations in neutral position, a dynamic study must be made and the adolescent must be referred to a specialist.

Another focus of care must be the moisturizing and integrity of the skin, which tends to remain dry and subject to infections. Daily moisturizing and the use of neutral soap to wash clothes is suggested.

Odontological monitoring must be done every semester, focusing on developing self-care in relation to oral hygiene.

Also in this age bracket, sleep apnea symptoms must be observed, and they include abnormal position in bed, waking up at night, nasal obstruction, snoring, and sleepiness during the day. The main cause of sleep apnea in adolescents with DS is obesity, for which a polysomnography is indicated. The quality and quantity of sleep is important in DS, since sleepiness during the day can generate mood changes, concentration problems, and decreased learning.[50,51]

Behavior changes in adolescence could mean depression or obsessive compulsive disorder and deserves monitoring by a specialist.

Another focus for this group is the prevention of physical and sexual abuse, through orientation and development of autonomy and self-care, besides orientation on the development of sexuality, as well as prevention of pregnancy and of sexually transmitted diseases. It is known that DS female adolescents are usually fertile and males, less frequently.

Ponderopostural monitoring follows the weight and height tables of Cronk.[41] The immunization calendar, in this age bracket, includes Hepatitis B, ADT vaccine (Adult Diphtheria and Tetanus), Yellow Fever, and MMR (Measles, Mumps, and Rubella) (Table 6).[47]

The therapy team for adolescents is composed of a psychologist, occupational therapist, physical educator, and nutritionist, also aided by an educator who acts with the psychologist as a tutor in schooling matters and preparation for

Table 5. Immunization calendar for children of four to eleven years old with Down Syndrome

Age	Vaccine	Dose
4 years	Trivalent Bacterial Vaccine (Diphtheria, Tetanus, and Pertussis)	2nd booster shot
	MMR Vaccine (Measles, Mumps, and Rubella)	2nd dose
	Varicella Vaccine*	2nd dose
10 years	Yellow Fever (regional recommendation)	One dose every ten years

Based on the National Immunization Program and nomenclature according to the Resolution of the Collegiate Board of Directors - RDC nº 61, of August 25, 2008 - National Agency for Sanitary Vigilance - ANVISA. Updated in 2012. * special vaccines that are not part of the calendar

Table 6. Immunization calendar for adolescents with Down Syndrome

Age	Vaccine	Dose
From 11 to 19 years old	Hepatitis B	1st dose
	Hepatitis B	2nd dose
	Hepatitis B	3rd dose
	ADT (Adult Diphtheria and Tetanus)	One dose every 10 years
	Yellow Fever	One dose every 10 years
	MMR (Measles, Mumps, and Rubella)	Two doses

Based on the National Immunization Program and nomenclature according to the Resolution of the Collegiate Board of Directors - RDC nº 61, of August 25, 2008 - National Agency for Sanitary Vigilance - ANVISA. Updated in 2012

Table 7. Immunization calendar for adults and the elderly with Down Syndrome

Age	Vaccine	Dose
From 20 to 59 years old	Hepatitis B (vulnerable groups)	Three doses
	ADT (Adult Diphtheria and Tetanus)	One dose every 10 years
	Yellow Fever	One dose every 10 years
	MMR (Measles, Mumps, and Rubella)	Single dose
60 years old or older	Hepatitis B (vulnerable groups)	Three doses
	Yellow Fever	One dose every 10 years
	Seasonal Influenza	Annual dose
	Pneumococcal 23 Valent	Single dose
	ADT (Adult Diphtheria and Tetanus)	One dose every 10 years

Nomenclature from the National Immunization Program and inserted nomenclature according to the Resolution of the Collegiate Board of Directors - RDC nº 61, of August 25, 2008 - National Agency for Sanitary Vigilance - ANVISA. Updated in 2012

the work market. The model for this age bracket is called *Down Adolescent*, with a weekly individual 30-minute session with each one of the professionals in the therapy team, and monitoring by a physician, nurse, social worker, and dentist, every semester or as demanded by the patient and his or her family.

The adolescents cared for by the Down Adolescent model participated in art experience and generation of income workshops.

The Autonomy Model is also offered to the adolescents, so as to develop competences for daily life and instrumental activities, and preparation for adult life. In this model there are two weekly consultations for the adolescent group and two for the parental group, with the duration of two hours, one hour with a psychologist and another hour with an occupational therapist. This activity is done in groups of six adolescents and their parents or caregivers who also receive orientation to continue the proposed activities at home.

Health care for adults and the elderly

Health care for adults and the elderly with DS must be focused on maintaining a healthy lifestyle (diet, body hygiene, sleep hygiene, and the practice of exercises), on the development of autonomy and self-care, on socialization, and on social and economic inclusion. In this phase questions of independence and future planning must be discussed with the family regarding the care and financial support of the person with DS. The questions of sexuality and prevention of pregnancy and sexually transmitted diseases continue being worked in this age bracket.

In this phase the following exams are needed annually: complete blood count, dosage of the Thyroid Stimulating Hormone (TSH) and of thyroid hormones (T3 and T4), in addition to auditory acuity every year, and visual acuity every three years.

In this phase of life, as well as in the previous phases, it is important to maintain the importance of cervical posture orientation and request radiological study of the cervical column in case of cervical pain, torticollis, weakness in the upper limbs, dizziness, or intestinal and vesical alterations. In this case a cervical radiography in the neutral position must be taken first. If there are no alterations it is possible to make a radiographic study in flexion and extension of the neck. In those cases with alterations in neutral position, a dynamic study must not be made and the patient must be referred to a spinal column specialist. In case of surgery under general anesthesia it is necessary to notify the anesthesiologist of the risk of hyperextending the cervical column during the anesthetic procedure and surgery itself.[6,8]

Odontological monitoring must be kept up annually.

In adults and the elderly sleep apnea is common, just as in other age brackets. The main cause of sleep apnea in the DS adult is obesity, and a polysomnography is indicated.

Behavior changes are more common in adult life for a person with DS and deserve special attention, for they could mean depression, obsessive-compulsive disorder, or mental deterioration by the increased risk of Alzheimer and early aging.

Another focus of care must be the moisturizing and integrity of the skin, which tends to remain dry and is subject to infections. Daily moisturizing and the use of neutral soap to wash clothes is suggested.

Females with DS must, as with other adults, follow an annual gynecological monitoring routine, as well as the males must follow a urological monitoring routine.

Ponderopostural monitoring follows the weight and height tables of Cronk.[41] The immunization calendar, in the adult bracket, includes Hepatitis B, ADT vaccine (Adult Diphtheria and Tetanus), Yellow Fever, and MMR (Measles, Mumps, and Rubella). The elderly also need the Pneumococcal 23 Valent and seasonal Influenza (Table 7).[33, 47]

The therapy team for adults is composed of a psychologist, occupational therapist, physical educator, and nutritionist, also aided by an educator who acts with the psychologist as a tutor in schooling matters and preparation for the work market. The model for this age bracket is called *Down Adult*, with weekly 30-minute individual sessions or in groups of six participants with each one of the professionals in the therapy team, and monitoring by a physician, nurse, social worker, and dentist, semi-annually or as demanded by the patient and his or her family. The adults in the Down Adult model participate in art experience, theater, and income generation workshops, in addition to physical conditioning groups.

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Summary of personal health care protocol for people with Down Syndrome

	New-born	Children from 1 to 10	Adolescents	Adults	Elderly
Exams	TSH (from 6 months to 1 year old)	TSH (annual)	TSH (annual)	TSH (annual)	TSH (annual)
	Complete blood count (at 6 months and at 1 year old)	Complete blood count (annual)	Complete blood count (annual)	Complete blood count (annual)	Complete blood count (annual)
	Karyotype*				
			Blood sugar, Triglycerides, and Lipidogram (in the case of obesity)	Blood sugar, Triglycerides, and Lipidogram (in the case of obesity)	Blood sugar, Triglycerides, and Lipidogram (in the case of obesity)
		Cervical column X-ray** (at 3 and at 10 years of age)	Cervical column X-ray** (IN)	Cervical column X-ray** (IN)	Cervical column X-ray** (IN)
	Echocardiogram***	Echocardiogram (IN)	Echocardiogram (IN)	Echocardiogram (IN)	Cardiological evaluation
Evaluations	Vision (6 months)	Vision (annual)	Vision (biannual)	Vision (triennial)	Vision (triennial)
	Hearing (6 months)	Hearing (annual)	Hearing (annual)	Hearing (annual)	Hearing (annual)
			Gynecological evaluation (annual)	Gynecological evaluation (annual)	Gynecological evaluation (annual)
Extra immunization		Varicella Hepatitis A			Seasonal Influenza
Therapy Team	Occupational Therapy	Occupational Therapy	Occupational Therapy	Occupational Therapy	Occupational Therapy
	Physical Therapy	Physical Therapy Physical Conditioning	Physical Conditioning	Physical Conditioning	Physical Conditioning
	Speech Therapy	Speech Therapy			
	Psychology	Psychology	Psychology	Psychology	Psychology
	Nursing	Nursing	Nursing	Nursing	Nursing
	Nutrition	Nutrition	Nutrition	Nutrition	Nutrition
	Social Work	Social Work	Social Work	Social Work	Social Work
	Physician	Physician	Physician	Physician	Physician
	Odontology	Odontology	Odontology	Odontology	Odontology
			Educator	Educator	
Guidelines	Positioning of the neck	Positioning of the neck	Positioning of the neck	Positioning of the neck	Positioning of the neck
	Global stimulation	Physical activity	Physical activity	Physical activity	Physical activity
	Stimulus to breast feeding	Healthy diet	Healthy diet - care with obesity	Healthy diet - care with obesity	Healthy diet - care with obesity
	Contact with other parents	Healthy life habits	Healthy life habits	Healthy life habits	Healthy life habits
	Community support	Socialization	Socialization	Socialization	Socialization
		Schooling	Schooling and curricular adaptation	Schooling and preparation for a job	Social and economical inclusion
		Stimulate self-care	Stimulate self-care and autonomy for the BDLA and IDLA	Stimulate self-care and autonomy for the BDLA and IDLA	Stimulate independence and inclusion in the work market, autonomy for the BDLA and IDLA
		Risk of sexual exploitation	Risk of sexual exploitation	Risk of sexual exploitation	Risk of sexual exploitation
		Behavior changes (autism)	Appropriate social behavior	Beware of signs of depression and obsessive-compulsive disorder	Beware of signs of Alzheimer, depression, and obsessive-compulsive disorder
		Risk of cervical lesion during leisure	Risk of cervical lesion by the use of computers, sports, and during leisure	Risk of cervical lesion by the use of computers, sports, and during leisure	Risk of cervical lesion by the use of computers, sports, and during leisure

Continuation...

	Attention to dry skin	Attention to dry skin	Attention to dry skin	Attention to dry skin
		Beware of sleep apnea	Beware of sleep apnea	Beware of sleep apnea
Beware of constipation	Beware of constipation	Beware of constipation	Beware of constipation	Beware of constipation
		Prevention of pregnancy	Prevention of pregnancy	Financial planning and future care

* The karyotype must be requested during the first year of life or at any time if it has not been done yet. ** Radiological evaluation must be done at 3 and again at 10 years old, and at any other times if there are symptoms (cervical pain, weakness, torticollis, vesical and intestinal alterations). When requested, it must be done first in the neutral position; if results are normal, proceed with the dynamic evaluation in extension and flexion. Some services in the care of DS choose clinical monitoring and do not ask for this exam. *** In case the first echocardiogram does not show any cardiac malformation it is not necessary to repeat it, and clinical monitoring is recommended. IN: If Necessary; BDLA: Basic Daily Life Activities; IDLA: Instrumental Daily Life Activities.

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