

THE EDSA ESSENTIALS ②

HEALTH CARE GUIDELINES FOR PEOPLE WITH DOWN SYNDROME



FOREWORD

Health Care Guidelines for People with Down Syndrome is an official document by the European Down Syndrome Association (EDSA) written by an international team of doctors and experts in Down syndrome under the direction of Prof. Alberto Rasore-Quartino.

People with Down syndrome frequently present alterations in the development, structure and functions of several organs or systems that can lead to complications – sometimes severe – for their physical health and deterioration in their quality of life.

Many of these complications can be prevented if they are kept in mind and treated at an early stage, with the correct application of a specific programme of truly preventive medicine.

If we accept, moreover, that good health is the fundamental basis for intellectual development, it is easy to conclude that full development and being able to make the most of all the capacities of each person with Down syndrome will depend to a large extent on their being in good health.

The programme presented by EDSA adapts to the health care standards of the majority of countries in Europe with a special focus on the Eastern European countries that have recently joined the European Union.

The programme has two parts: a set of current recommendations, understood as a general guide for exploration, in relation to each age and before specific symptoms of Down syndrome, and a justification of why special attention is paid to certain systems and why specific evaluations are recommended.

EDSA would like to thank Prof. Rasore-Quartino and his collaborators for their work and hopes that its application may help many doctors to care for and improve the health of people with Down syndrome.

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HEALTH CARE GUIDELINES FOR PEOPLE WITH DOWN SYNDROME

European Down Syndrome Association (EDSA)

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People with Down Syndrome (DS) need the usual health care procedures provided for everyone.

However, they have increased risks of congenital malformation and during their lives they may develop certain medical problems occurring at a much higher frequency than in people without the syndrome. This requires a medical programme that can provide both rehabilitation programs and social inclusion. Medical intervention has the aim of preventing or treating in due time diseases that may be the cause of increased morbidity and mortality, if neglected. It is therefore necessary to know the natural history of DS, the medical complications occurring and their prevalence in different age-groups.

For a long time, scientists everywhere have been preparing health care guidelines, from the USA to Europe (Italy, France, Spain, UK, Ireland, Belgium, the Netherlands, Germany, etc.).

EDSA proposes special health care guidelines which could be useful for all paediatricians and general practitioners caring for people with DS of any age. These guidelines are based on previous publications and on the personal experience of the authors, trying to treat the most important medical aspects and to offer complete but simple and comprehensible recommendations, which can be useful for everyone.

Since these guidelines are based on current knowledge, they should be modified as new information becomes available.

We would also like to stress that these procedures have different finalities in the child and the adult. In the former, they are aimed at the optimisation of the rehabilitation process, while in the latter they are needed to maintain acquired abilities.

They also include special recommendations for the educational and developmental needs of people with DS, according to modern concerns for their well-being (*sentences written in italics*).

Ultimately, they are intended to give them a full and meaningful life.

The prenatal period (1).

General information on DS is easily available for individuals, parents and families. The paediatrician, geneticist or obstetrician can be asked to counsel a couple about the risks of conceiving a foetus with DS.

The main topics discussed with these people are:

The approach to the available technical procedures for prenatal diagnosis

The available treatments

The options for bringing up a child with DS

Assistance in the decision making process (non directive approach)

(1) It is well known that the prenatal period is of paramount importance for the preparation of the couple with respect to the future life of the unborn child. Complete information on the risks of malformations, genetic diseases, the possibilities of prenatal diagnosis and on the alternative decisions to be taken (genetic counselling) should be given from qualified professionals (paediatricians, geneticists, obstetricians, etc.). It must be stressed that the approach to the decisional process is non-directive, respecting the personality and beliefs of the individuals in question.

Health care guidelines for the newborn (birth to 1 m).

Clinical diagnosis and its communication to parents (2)

Cytogenetic analysis

Clinical and neurological assessment

Clinical and instrumental investigations for congenital malformations (echocardiography, abdominal echography, etc.) (3)

Ophthalmologic examination (4)

Auditory screening (otoacoustic emissions) (4)

Blood tests for polycytemia, leukemoid reaction, leukaemia, etc (5)

Routine neonatal screening

Encouragement of breast-feeding

Parental psychological support

(2) Clinical diagnosis at birth is possible in most cases. Difficulties can arise in very small babies, as is the case with premature or small-for-age newborns, or if there are severe clinical problems diverting the attention from the phenotypic

characteristics of the infant. The diagnosis, even if only dubious, should be communicated altogether to both parents as early as possible, even if there is no result from chromosome analysis yet, in a simple and concise way, explaining the physical characteristics of the newborn and pointing out the positive aspects of the future development, the learning abilities and the possibilities of independent life. In general, more than one session with the family is necessary. It is also desirable to offer genetic counselling and psychological support.

(3) In DS, congenital malformations can be numerous and therefore must be correctly recognised in order to treat them properly and in time, with the aim of avoiding complications that can sometimes be very severe (Hall, 1988). Congenital heart disease is the most common severe malformation: about 50% of newborns are affected. They represent 7% of all infants with congenital heart defects. Endocardial cushion defect, or atrioventricular canal defect, is the prevalent anomaly, averaging nearly half of the total. An early diagnosis is desirable, since most of the anomalies are treatable by effective surgery. Cardiac defects with increased pulmonary flow are most frequent; affected infants become symptomatic at an early age, developing pulmonary artery hypertension, cardiomegaly, cirrhosis of the liver and congestive heart failure. Patients show growth retardation and recurrent respiratory infections, resulting in high morbidity and mortality. Pulmonary vascular obstructive disease is a severe complication, the development of which generally prevents surgical correction. Since pulmonary artery hypertension and pulmonary vascular disease in DS develop earlier than in children without DS, surgical management is recommended as early as possible. Moreover, surgical mortality has dramatically decreased in recent years and the long-term

prognosis is good (Marino and Pueschel, 1996). Other congenital malformations, although rare, should be thoroughly investigated, since surgical repair is almost always possible. Among gastrointestinal anomalies, duodenal stenosis, which is found in 4-7% of newborns with DS, represents 30-50% of all duodenal stenoses. The relatively high incidence of congenital megacolon has to be remembered (3.4% versus 0.02% in normal children). Pancreas annular and anal imperforation are also relatively frequent.

- (4) Sensory defects, when present, have great importance in the general pattern of mental development of children with DS, since they can significantly reduce the efficacy of any rehabilitation program, even the most rigorous one, in the acquisition of new abilities. This is particularly true in the first year of life. Ocular abnormalities are definitely more common than in other children. From a practical point of view, it is necessary to point out the clinical significance of strabismus and of refractory defects, which can hinder correct vision and hence add an organic defect to the underlying mental disability. An early diagnosis is essential, in order to correct them as early as possible. Surgical correction should also be provided, if necessary. It is well known that even very young children do not have difficulties in wearing spectacles, if they receive a real benefit. Cataract is another excessively frequent ocular defect observed both in newborns and adults. When children with DS have some sort of hearing abnormality, they cannot express the ability of employing the complex strategies necessary to compensate for their deficiency. Conflicting data on the prevalence of hearing abnormalities in DS is available. Evidence is given of an excess of middle ear pathology. A characteristic serous otitis can develop in

the first years of life and can often persist through adulthood. In fact, all the proposed treatments for such ear infections have a low success rate and hearing problems are the consequence of delayed recovery. About 80% of persons with DS of any age have a more or less severe hearing deficiency, mostly a conductive one. A preventive approach to the hearing problems of children with DS seems therefore of the utmost importance, in order to help them acquire good communicative ability and a satisfying socialisation.

- (5) In DS newborns inefficient regulation of blood cell production is usual (Weinstein, 1978), leading to various haematological abnormalities such as polycytemia (which should be treated, in order to avoid cerebral damage), thrombocytopenia, thrombocytosis, and a higher or lower leukocyte count. These abnormalities are time-limited and are the consequence of a defective control in the production of haemopoietic cells in one or more cell lines (Miller and Cosgriff, 1983). The most severe aspect of defective haemopoiesis is leukaemia. In DS the risk of developing leukaemia is 10 to 20 times higher than in normal children (Rosner and Lee, 1972). Moreover, in DS 25% of all leukaemias are evident at birth; 15% of the congenital leukaemias develop in newborns with DS. The response to treatment, prognosis and other characteristics are similar. The abnormal sensitivity to methotrexate found in children with DS has been attributed to the prolonged clearance of the drug in these persons (Garré et al, 1987). In 17% of infants with DS a form of acute, transitory leukaemia can develop. Its clinical and haematological features are indistinguishable from those of common, acute leukaemia, except for its course leading to spontaneous and complete remission. A differential diagnosis is usually very difficult and severe problems can arise for therapeutic decisions (Cominetti and Rasore-Quartino, 1988).

Health care guidelines in the first year (1 to 12 m).

Periodic clinical and neurological assessments	every 2 months
Evaluation of appropriate food intake and of normal growth for DS (6)	every 2 months
Echocardiography (if not done at birth)	
Visits and screening for visual and hearing defects	at 6 and 12 months
Blood tests for thyroid disease, anaemia, celiac disease, immunological defects, etc. (7)	at 12 months
Vaccinations according to local schedules	

Refer to parents associations or to DS support groups

Refer to Centres for early rehabilitation

Periodic family psychological support with special emphasis on siblings

- (6) The importance of a well-balanced diet should be emphasized, as is weaning in due time. Auxologic controls are fundamental, since linear growth retardation is characteristic of DS. Stature is generally stabilized at minus 2-3 standard deviations on normal growth charts. Special growth charts for children with DS are also available (Cronk et al, 1988; Myrolyd et al, 2002). The mechanisms responsible for short stature have not yet been completely explained, but multiple causes, such as malabsorption, congenital heart disease and hypothyroidism may be responsible. A great deal of interest was focused on the role of growth hormone (GH) and of somatomedins/insulin-like growth factors (IGF), since these hormones are not only essential for body growth, but also for the development and maintenance of the nervous system. Low levels

of IGF-I have been found in children with DS (Rasore-Quartino et al., 1987). Most authors, on the contrary, found a normal GH secretion. This notwithstanding, GH treatment has been proposed for children with DS and impaired growth, irrespective of their GH and IGF-I levels. Interesting results have been obtained with acceleration of growth velocity (Annerén et al., 2000). Nevertheless, at present, the role of the therapy is still controversial due to the lack of long-term results and for the possible complications (hypertension, hyperglycaemia). GH treatment is therefore not recommended as a standard therapy in children with DS.

- (7) Since the first descriptions of DS and for almost a century, hypothyroidism was considered a constant feature of the syndrome. Only when laboratory tests for thyroid function became available, did evidence come up that most people with DS are actually euthyroid. Nevertheless, it was also demonstrated that a higher incidence of thyroid disorders, chiefly hypothyroidism, is characteristic of DS. According to the literature, congenital hypothyroidism in DS varies from 0.7% to 0.10%, while in non-trisomic newborns it varies from 0.015% to 0.020%. Figures for acquired hypothyroidism are also highly variable (from 13% to 54% in DS, versus 0.8% to 1.1% in the normal population. Fort et al., 1984). Two forms of hypothyroidism can be distinguished. The most frequent, the so called compensated hypothyroidism, shows only increased levels of thyroid stimulating hormone (TSH), while the levels of thyroid hormones (T3 and T4) are within normal limits. Increased TSH represents a central response to the reduction of functional thyroid tissue on an immunological level and is followed by a progressive decrease of T3 and T4 values. Although this is commonly the course of the disease, in DS, TSH levels often fluctuate without any modification of thyroid function. These transient thyroid neuroregulatory dysfunctions are possibly related to inappropriate secretions of TSH or to

reduced sensitivity to TSH itself. An increased frequency of antithyroid antibodies is also found. Some authors claim that significantly lower IQs are found in persons with DS and elevated TSH levels. Frequently in DS, hypothyroidism is the consequence of an autoimmune disorder (Karlsson et al., 1998). Initially, only increased TSH levels are detected, and then the hormone deficiency develops, showing reduced T3 and T4 dosage. As the disease progresses, clinical symptoms appear. Unfortunately, they may not be recognised or can be mistaken for the features of the syndrome itself (dullness, increased fatigability, loss of attention, etc.), mainly in adolescents and adults, when some neurological or psychiatric symptoms may more easily appear, irrespective of thyroid status. Since untreated hypothyroidism can interfere with normal neuronal function, causing decreased intellectual abilities, appropriate substitutive therapy is strongly recommended. Personal investigations confirm that persons with DS have an increased risk of developing hypothyroidism at any age. One person out of 12 has either compensated or clinical hypothyroidism (Rasore-Quartino and Cominetti, 1994).

Intestinal malabsorption in DS is responsible for intestinal disturbances and for growth retardation in some children. Celiac disease, or gluten intolerance, in particular, has an increased prevalence in these persons. Actually, while its prevalence in the normal population is 0.43%, in DS it is 6% (Bonamico, Rasore-Quartino et al., 2001). Gluten is a component of wheat, rye, barley and other cereals. The disease, in its typical, rather uncommon form, is very severe: it usually develops in early childhood, after the introduction of gluten into the diet (approximately at 6 months). It manifests itself with diarrhoea, bulky stools, prominent abdomen, and poor thriving. At present, more frequent, moderate or atypical forms are found, appearing late in childhood or in adolescence and

showing scarce or absent intestinal symptoms, hypovitaminosis, anaemia, and stunted growth. Silent forms are also observed. The pathogenesis of celiac disease is still controversial. Recent studies ascribe the responsibility of the mucosal damage to an abnormal immune response to gliadin (Marsh, 1992). The diagnosis is based on intestinal peroral biopsy that shows the typical histological lesions of the mucosa, represented by a partial or total villous atrophy. IgG and IgA gliadin antibodies are a reliable and simple screening test, useful for detecting subjects who are eligible for intestinal biopsies. In DS IgAAGAs show an excess of positive results not confirmed by bioptic data: IgGAGAs, less specific, but more sensitive, are even more often positive (Storm, 1990). A more reliable, highly specific screening test is the antiendomysium immunofluorescence test, now almost universally substituting gliadin antibodies. Antitransglutaminase antibodies are also both specific and sensitive. The mainstream treatment for celiac disease is the gluten-free diet, which brings a complete recovery. A strong commitment and constant surveillance are required with patients, as compliance is often difficult to obtain.

Health care guidelines from 1 to 6 years

Clinical and neurological assessment	twice a year
Periodic assessments of diet and prevention of obesity (8)	
Periodic growth assessment	
Dental examinations (9)	once a year
Orthopaedic examinations (X-rays for atlantoaxial instability / subluxation, if necessary, etc.) (10)	once a year
Visits and tests for visual and hearing defects	at 3 and 6 years
Blood tests for thyroid disease,	

anaemia, celiac disease, autoimmune disorders, etc. once a year

ORL controls for hypertrophic tonsils and adenoids,
for mechanical respiratory problems, sleep apnoea, otitis, etc. once a year

Vaccinations according to local programs

Specific vaccinations (influenza, pneumococcus, etc.) if at risk

Rehabilitation programmes (continuation)

Logopedic support

Social inclusion (maternal school)

(8) The tendency to obesity of people with DS is well known. It manifests itself mainly in young adults: therefore we must establish a correct prevention from childhood, with particular attention to adolescence. We should intervene in nutrition and in physical activity. The assessment of caloric intake should be a constant rule. Excess of glucose- and lipid-rich food should be avoided, protein rich food should be preferred, but in a well balanced diet.

(9) Dental anomalies are a common problem, the solution to which is not an easy task. Moreover, objective difficulties found in visiting and specifically treating children and adults with intellectual disabilities lead to underestimation of actual buccal disorders. A peculiar oral and dental anatomy, developmental anomalies and malocclusion are frequent. On the contrary, caries seems to be less frequent than in normal persons. If oral hygiene is poor, gingivitis and periodontal disease are likely to occur, leading to early and total tooth loss. Dental checks should be constant from childhood and for the whole life. Accurate orthodontic help should be available, in order to avoid the depressing consequences of dental decay (Lowe, 1990).

(10) Muscular and orthopaedic anomalies are well known in DS. Muscular hypotonia and joint hyperlaxity are almost constant. Flat foot, genu valgum, patella instability are main causes of walking problems, even of severe static troubles such as scoliosis and cyphosis. Prevention is necessary and is achievable through early and correct mobilisation, and an active life associated with sport activities. The clinical significance of atlantoaxial instability received specific attention in the last years. Its prevalence is elevated in DS (10-15%) and is usually asymptomatic (Pueschel and Scola, 1987; Pueschel, Scola and Pezzullo, 1992). An increased risk of dislocation exists after cervical traumas or abrupt head movements, with neurological complications occurring by cervical cord compression. Dislocation can produce quadriplegia with incontinence or paraplegia that may have a sudden onset or can be preceded by head tilt, abnormal staggering gait and the emergence of neurological signs. Diagnosis is confirmed by X rays that demonstrate a distance superior to 5 mm between the anterior side of the odontoid process and the posterior margin of the anterior arch of the atlas. Magnetic Resonance Imaging or Computerized Tomographic Scans are also useful tools for diagnostic purposes. Children and youngsters at risk should not be allowed to practise somersaulting, trampolining, boxing and fighting or similar sport activities (Cremers et al, 1993). For symptomatic cases, vertebral fusion is the preferred surgical procedure (Aicardi, 1992).

Health care guidelines from 7 to 12 years

Clinical and neuropsychiatric assessment	annually
Dental examinations	annually
Periodic assessment of diet and prevention of obesity	

Growth assessment	annually
Ophthalmologic examinations	annually
Audiologic examinations	annually
Orthopaedic examinations	annually
ORL examinations for hypertrophic tonsils, mechanical respiratory problems, sleep apnoea, otitis media, etc	annually
Blood tests for thyroid disease, celiac disease, leukaemia, autoimmune disorders, immunological deficiencies, etc.	annually
Assessment of sexual development and preparation of the family to cope with this (11)	
<i>Family psychological support</i>	
<i>School insertion and recreational activities</i>	

(11) Sexual maturation is similar to that of the general population. In males, testicular volume and penis dimension reach normal values during puberty. Cryptorchidism is common and should be corrected early in life, due to the risk of malignant degeneration in adulthood. In females, the development of secondary sexual characteristics follows a regular pattern. Menses are regular. Menopause occurs early. Fertility is reduced in females: only a small number of pregnancies have been observed, resulting in both normal and trisomic babies. Males are almost invariably sterile (only a few cases of fatherhood having been reported).

Since insertion into society is becoming more and more frequent, adolescents should be prepared for a sexually active life. It is the task of parents, psychologists and physicians to patiently and delicately teach young people with DS about the possibilities that life offers them, together with the possible

associated dangers. Periodic gynaecological visits are recommended as for normal women. Contraception can be provided in particular cases.

Health care guidelines from adolescence to early adulthood

Clinical assessment	annually
Neurological and psychiatric assessment (depression, autism, etc.)	annually
Periodic assessment of dietary habits	
Prevention or correction of obesity	
Ophthalmologic examinations	annually
Audiologic examinations	annually
Dental examinations	annually
Orthopaedic examinations	annually
Blood tests for thyroid disease, celiac disease, autoimmune disease, immunological deficiencies, etc.	annually
Assessment of sexual development	
Gynaecologic examinations	
Evaluation of need for contraception (in sexually active females)	
<i>Monitor school progress</i>	
<i>Encourage recreational activities</i>	
<i>Stimulate sport activities</i>	
<i>Evaluate the possibility of active employment</i>	
<i>Envisage possibilities of independent life</i>	
<i>Psychological family and individual support</i>	

Health care guidelines in adulthood and old age.

(periodicity should always be established according to individual needs)

Clinical assessments

Assessment of dietary habits

Prevention or correction of obesity

Cardiologic examinations (echocardiography for aortic regurgitation or mitral valve prolapse) (12)

Neurological and psychiatric assessments (early ageing, depression, Alzheimer's disease, autism) (13)

Orthopaedic controls

Gynaecologic examinations

Dental examinations

Ophthalmologic examinations

Audiologic examinations

Blood tests for anaemia, thyroid disease, celiac disease, autoimmune disorders, etc.

Clinical and instrumental checks for oncologic diseases

Specific vaccinations for age or if at risk (influenza, pneumococcal, etc)

Programs for enhancement/maintenance of acquired abilities

Stimulate physical activity

Encourage recreational activities

Individual psychological support

Envisage necessity of living in community in foster homes, protected homes, or of admission to institutions

(12) Available data on cardiac status of adults with DS is not as numerous as for children. From recent literature, it is evident that they may also have heart problems other than congenital defects. The most frequent anomalies found in asymptomatic adults are mitral valve prolapse and aortic regurgitation, with a prevalence of up to 70%. These defects seem to occur only in adults, since they have never been detected in childhood (Goldhaber et al., 1987; Marino and Pueschel, 1996). Obviously, accurate diagnostic investigation is recommended in young adults, especially before dental and surgical procedures, in order to detect these defects. Antibiotic prophylaxis for endocarditis should also be taken into consideration.

(13) Impaired mental ability and delayed psychomotor development are constant in DS and show a wide range of ultimate attainment that can to some extent be positively influenced by present educational strategies. Neuropsychiatric problems become prevalent with age, including seizures. In adults, there is a constant, though variable, decline in intelligence. A reduction in thought elaboration ability, in particular for abstract thought and logical performance is likely to occur with advancing age, but earlier than in normal people. Dementia is also characteristic of ageing in DS, showing striking similarities to Alzheimer's disease and

appearing in a number of subjects over the age of 50. Clinically, affected individuals show deterioration of mental and emotional response, apathy or excitement, irritability, temper tantrums, loss of previously acquired vocabulary and a decline in personal cleanliness. The progression is often very rapid. Seizures can be an early sign of Alzheimer's disease. No effective therapy is known at present. Recent investigations suggest that most adults with DS undergo normal, albeit probably early ageing and that they may be at lower risk of Alzheimer's disease than previously supposed (Devenny et al. 1996). It is also possible that early intensive rehabilitation and social inclusion are beneficial in slowing down mental deterioration and ageing.

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WHAT IS EDSA?

EDSA is a non-profit international Association whose goals are:

- to promote all actions and efforts which contribute to the welfare of persons with Down Syndrome according to the rights awarded to them by their own constitution, the Declaration of Human Rights and rights of persons with disabilities by the United Nations, and the European convention relating to the protection of fundamental rights and liberties.
- Promote the well-being of persons with Down Syndrome in every possible way and area, and in every aspect of life: health, education, personality, autonomy and integration in the society according to the individual's aspirations and capabilities.
- To encourage all scientific efforts towards advances and improvements in medical care, education, rehabilitation, vocational training, employment, leisure and independent living.
- To further the interests of persons with Down Syndrome by securing all necessary resources, support and services to that end.
- To create a bond among all persons with Down Syndrome, their families, friends and associations.

Article 3. EDSA BY LAWS



EDSA JOURNALS:

- Down Syndrome News and Update
- Journal de la Trisomie 21

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