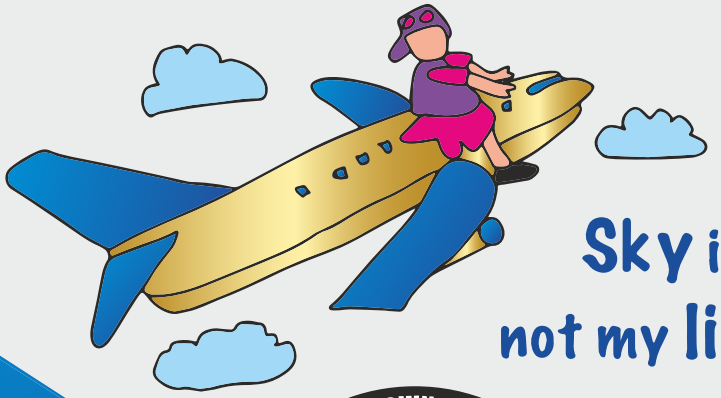

Sky is not my limit

INDIA INTERNATIONAL DOWN SYNDROME CONFERENCE

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DOWN SYNDROME
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INDIA INTERNATIONAL DOWN SYNDROME CONFERENCE 2023

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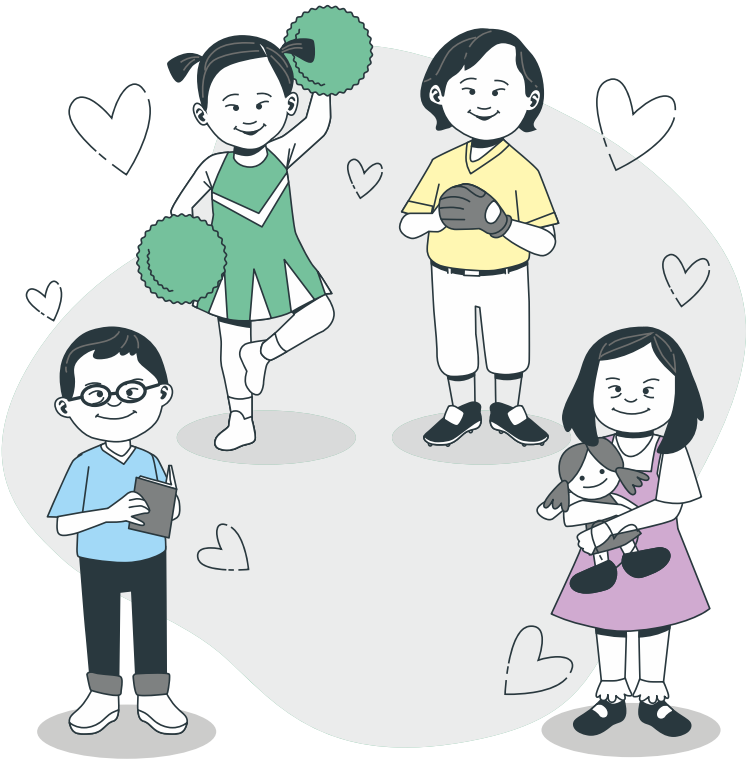
Index

MANAGEMENT OF CHILDREN WITH DOWN SYNDROME

Down syndrome (DS) is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. Majority of mothers of babies with DS in developing countries are in the 20s, as pregnancy above 35 years is comparatively less in these countries [1]. The management of DS has evolved from a purely medical model to a social, and now, a rights model. Regular and repeated counselling of parents is of utmost importance to ensure a sustained management program [2]. Awareness and educational programs for the public at large are important to create an inclusive society by which these children are able to lead an independent life.



OBJECTIVES OF THE GUIDELINES

The objective of these guidelines is to detail briefly about the involvement of the various systems, and formulate recommendations for management. To provide a uniform structured guideline for the management of children with DS, that can be practiced across the country. To recommend early and sustained interventional therapies to enable children with DS lead a healthy and independent life.

Growth potential of children with DS is less compared to children with normal karyotype. Both height and head circumference are affected. Final height is less in comparison with that of individuals of same sex with normal karyotype. Growth is more compromised in children with DS who have major congenital heart diseases or other systemic diseases.

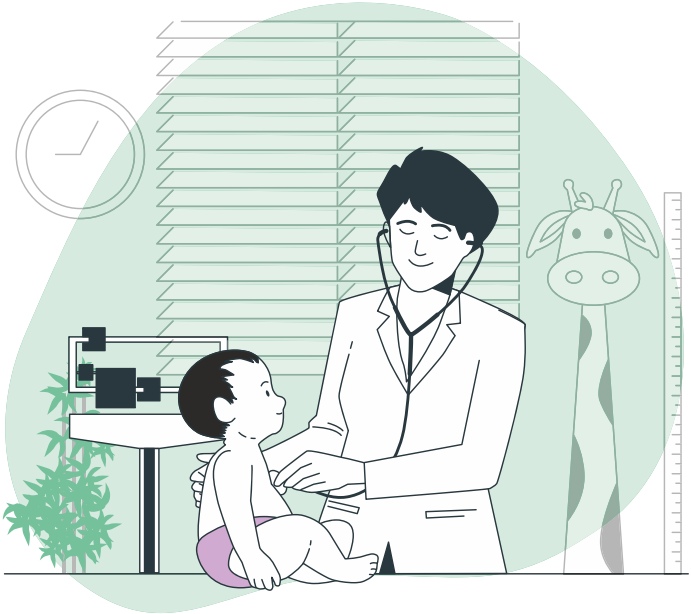
DS specific growth charts are to be used for monitoring growth of children with DS and interpretation of growth faltering. Height/length, weight and head circumference must be documented at birth, during immunization visits at 6, 10 and 14 weeks, at 6 months, 1 year and 6 monthly thereafter.

Children with DS tend to become obese, especially during adolescence. This has to be anticipated and measures should be taken for prevention. Likely determinants of obesity include increased leptin (a hormone released from adipose tissue containing the obese gene), decreased resting energy expenditure, comorbidities, depression, unfavorable diet and low physical activity. Obesity is associated with obstructive sleep apnea, dyslipidemia, hyperinsulinemia, and gait disorder

Prevention and management of obesity is similar to that for all children, and includes appropriate exercise programs and dietary modification.

Recommendations

The height/length, weight and head circumference should be regularly monitored and plotted in a DS specific growth chart. Till we have an Indian Growth chart for children with DS, we can use any growth chart available. It is the growth pattern that is important and not the percentile shown in the chart as our children would be shorter.



The high prevalence of congenital heart disease (CHD) in DS (40-60%) together with poor sensitivity and specificity of clinical examination, justifies routine ECG screening at the earliest opportunity in these babies. The cardiac status should be established by 6 weeks of age.

Most cardiac issues in DS are correctable through timely intervention and their management should be on the same lines as for children without DS. It is important not to delay correction in conditions such as atrio-ventricular septal defect (AVSD), ventricular septal defect (VSD), and patent ductus arteriosus (PDA), because of the propensity for rapid development of pulmonary vascular disease. The presence of DS does not significantly increase surgical risk. However, for complex defects such as unbalanced AVSD, and tetralogy of Fallot (TOF) with AVSD, management and outcome need to be carefully individualized.

Recommendations

I) Routine screening echocardiogram should be done at birth and cardiac status established by 6 weeks of age.
ii) There should be no delay in correction of large shunt lesions as it can lead onto pulmonary vascular disease.

Thyroid disorders are common in children with DS (24-50%). Both hypothyroidism and hyperthyroidism are described, but hypothyroidism is much more common and includes subclinical hypothyroidism (SCH), congenital hypothyroidism (CH), and autoimmune thyroid diseases (ATD).

SCH is the most common thyroid abnormality in children with DS. Almost 50% of children with SCH in DS have thyroid antibodies and half of these may progress to overt hypothyroidism. Congenital hypothyroidism has been reported to be about 28–30 times more common in neonates with DS. The TSH and T4 cutoffs used for screening CH in children with DS are the same as that for normal children.

Children with DS have short stature; even though, there is no overt growth hormone (GH) deficiency. However, selective Insulin like growth factor-1 deficiency has been reported in children with DS older than 2 years. No benefit in final adult height has been demonstrated with GH therapy. Pubertal children often show (high luteinizing hormone, LH; high and follicle stimulating hormone, FSH), which is progressive from birth to adolescence. Pubertal progression is also often delayed. Low bone mineral density is common and is due to low physical activity, less sun exposure, obesity, decreased muscle mass, associated illnesses (celiac disease) and drugs.

Recommendations

I) CH is not always picked up in the newborn screening, hence repeat screening at 6 months is recommended. Screening has to be repeated at the age of one year, and every year thereafter.
ii) Current recommendation for SCH is to treat only when the TSH remains persistently above 10 mIU/mL, or if there are clinical symptoms of hypothyroidism or goiter, or if thyroid autoantibodies are present.

HEARING ISSUES

It is estimated that over 50% of people with DS have hearing impairment, which can range from mild to profound. Early hearing loss has a significant impact on speech and language development of children with DS, resulting in disproportionately severe speech delay. Orofacial and craniofacial maldevelopment associated with DS contributes to inner, middle and outer ear problems. Large adenoids, small nasopharynx, impaired swallowing and narrow horizontal eustachian tubes are contributory factors. There is also a higher incidence of chronic rhinosinusitis and adenoid hypertrophy in these children. Cochlear implant is reported to be successful in these children for the management of severe sensorineural hearing loss.

Recommendations

I) Neonatal screening is recommended followed by a full audiological assessment between 6 and 10 months; and intervention initiated if needed. An audiological review should be carried out at around 18 months in a manner appropriate for the cognitive level, which should be repeated at least yearly until age 5 and thereafter 2 yearly for life.
ii) Regular de-waxing is advised for wax accumulation.
iii) For otitis media with effusion - grommet insertion is advisable, with adenoidectomy in case of recurrent issues.
iv) Sleep studies may be done in the presence of suggestive symptoms, where available, as there is a high prevalence (50-70%) of obstructive sleep apnea (OSA).

BLOOD RELATED DISORDERS

Nutritional anemia: Many children with DS in developing countries do have anemia, which in turn can affect the cognitive status. Hence, yearly blood counts must be done at least till 5 years of age.

Recommendations

I) All newborns with DS should have a complete blood count (CBC) and peripheral smear examination (PBS), which should be repeated yearly till 5 years.
ii) All children with previous TL-DS should be monitored for progression to Myeloid Leukemia-Down Syndrome (ML-DS) with 3 monthly clinical review and CBC and PBS until the age of 2 years. If the CBC and PBS are normal and there are no clinical features of ML-DS, monitoring should continue 6-monthly till the age of 4 years.

EYE-RELATED PROBLEMS

Ocular disorders have a high prevalence (60%) among people with DS. Nasolacrimal duct obstruction is seen in 10-36% of infants, compared with 2-4% of infants with normal karyotype. Refractive errors occur at an early age and are about 10 times more common. Around 54% of children with DS require prescription spectacles in preschool. Around one third children with DS will have visual defects by 18 months to 2 years, and around 50% are likely to have refractive errors by the age of 4 years. Congenital cataract as well as developmental cataract, nystagmus, and congenital glaucoma are also more prevalent. Various studies have shown that difficulty in accommodation in DS can be corrected using bi-focal lenses. Strabismus occurs in 25-30% children with DS, compared to 2-4% children without DS. Keratoconus is also seen in up to 15% children with DS, and regular monitoring is needed to avoid visual loss. Amblyopia is seen in up to 20%children with DS.

Recommendations

I) All newborns with DS should have an eye examination at 4-6 weeks to exclude congenital glaucoma, cataract and other eye abnormalities. Visual behavior needs to be monitored by a pediatrician before their first formal ophthalmological review.
ii) Between the age of 18 months and 2 years, all children should have an ophthalmological review, which is to be repeated at 4 years and thereafter every 2 years throughout life.
iii) Any child/adult with pain, and/or changing vision, or visual disturbances/red eye should be referred for urgent ophthalmological opinion.

SKIN RELATED ISSUES

Dry Skin and excessive thickening of the skin on palms and soles occur more frequently in individuals with DS. Dry skin predisposes individuals to irritant as well as allergic contact dermatitis. Acanthosis nigricans (a velvety, darkening of the skin in hidden areas) is also noted and is a marker of insulin resistance, and associated with obesity. Alopecia areata affects 6–10% of those with DS and is associated with autoimmune conditions such as vitiligo and hypothyroidism. Pustules, nodules and abscesses in the arm pits and groin are common. The response to treatment is often poor. Accelerated ageing is considered part of the DS phenotype and the signs include greying, thinning of hair, skin atrophy, early development of rhytids (wrinkles), and lentigines [28].

Recommendations

A thorough cutaneous examination, with particular attention to the commonly affected locations like scalp, axilla, groin, and feet should be done at all routine clinical visits.

MUSCULOSKELETAL ISSUES

Musculoskeletal problems in DS are due to central hypotonia, laxity of ligaments and excessive joint mobility. Autoimmune conditions and endocrine issues like hypothyroidism and osteoporosis further add to these problems. Foot, hip and spine are common areas involved in cases of DS, resulting in reduced mobility, lower bone density and high risk of injuries. Cervical spine instability due to issues with first and second cervical vertebrae result in neck pain, abnormal head/neck posture and motor symptoms in limbs. Upper cervical spine instability has the most potential for morbidity and, consequently, requires close monitoring. Rarely, spinal compression may lead to sudden death. Plain radiographs do not predict well the increased risk of developing spine problems, hence routine radiologic evaluation of the cervical spine in asymptomatic children is not recommended. Any child who has significant neck pain, radicular pain, weakness, spasticity or change in tone, gait difficulties, hyperreflexia (overactive reflex), change in bowel or bladder function, or other signs or symptoms of myelopathy must undergo plain cervical spine radiography in the neutral position, and referred, as required.

Recommendations

I) Hip and foot examination should be done in the first year of life.
ii) All children with DS and a limp should be evaluated with a hip X-ray.
iii) Routine screening of cervical spine is not recommended, unless they are engaged in contact sports, have any symptom of atlanto-axial instability or as a part of pre-anesthesia checkup.
iv) Scoliosis screening should be considered in the presence of suggestive findings.

RENAL AND GENITOURINARY INVOLVEMENT

Congenital anomalies of kidney and urinary tract (CAKUT) are noted in DS . With increased survival, a large number of these patients present with chronic renal failure.

Recommendations

I) A thorough physical examination to identify anomalies such as hypospadias, cryptorchidism, testicular cancer, and kidney malformations should be done. However, routine screening for renal and urological problems is not recommended.
ii) Postnatal follow up need to be done to rule out any renal problems, if anomaly scan had showed any abnormality.
iii) Yearly testicular examination should be done.

ORAL HEALTH ISSUES

Relatively large and fissured tongue with small oral cavity, malocclusion, mal-alignment, dental agenesis, enamel hypoplasia, delayed dentition, dental caries and periodontal disease are the common oral health issues encountered . The first tooth eruption is usually at the age of 12 to 14 months and may be completed only by 4 to 5 years of age. Permanent teeth eruption may also be delayed. Orthodontics (braces) may improve some of the issues associated with the mal-alignment, but it could interfere with speech and many find it difficult to tolerate.

Recommendations

I) First dental visit should be within 6 months of the first tooth eruption or by 1 year of age; and thereafter yearly till 5 years of age.
ii) Brushing should be done twice daily with a soft toothbrush and fluoride toothpaste.
iii) Preferably floss daily, even if there is gum bleed, as brushing and flossing help to keep the gums clean and minimize inflammation.
iv) Limit the amount and frequency of sugar and refined foods.

Gastrointestinal Manifestations

Structural and functional disorders of gastrointestinal tract seen in DS include esophageal atresia/tracheoesophageal fistula, duodenal/jejunal atresia or stenosis, annular pancreas, anorectal anomaly and Hirschsprung disease. Feeding difficulty (due to poor oromotor function), gastroesophageal reflux, constipation, toddler diarrhoea, and gall stones are other disorders noted. Celiac disease is seen in 5% of those with DS.

Recommendations

I) Complete physical examination of the infant including inspection of the perineum for anal patency.
ii) Enquire about passage of meconium
iii) Growth monitoring using DS growth charts as a screen for malabsorption; consider testing for coeliac disease, if symptomatic

Various studies have shown that adults with DS have hypergonadotropic hypogonadism (higher levels of FSH and/or LH). This is due to cell dysfunction of certain cells. Despite the gonadal dysfunction, puberty occurs on time and progresses at a typical rate as in other children. This emphasizes the need to counsel the caregivers to prepare children for upcoming pubertal changes.

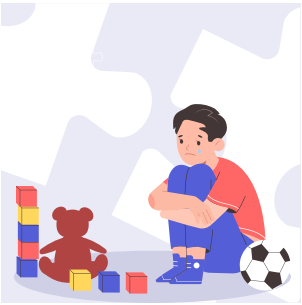
Sexuality: In the past, sexuality was not considered an issue for young people with DS because of the inaccurate belief that intellectual impairment was equivalent to a state of permanent childhood. In fact, all individuals with DS do have intimacy needs and sexual feelings as in other children and should be counselled accordingly. It is important to recognize these for planning education, housing and other programs.

Fertility: Men with DS have lower fertility, but 70% of women with DS are fertile. However, there is an increased chance of miscarriage. There is a 50% chance of having a child with DS, and a higher chance of having other congenital anomalies. Contraceptive advice should be provided by professionals with skills to support people with DS .

Management of menstrual cycles: Girls should be educated about menstruation in advance, so that they know whom to approach and what needs to be done. Most girls will be able to cope independently with their periods but they initially may need some support. Simple measures to treat dysmenorrhea should be taught to the caregivers. They may also experience premenstrual symptoms, but may not be able to report these symptoms. Hence, parents/caregivers should be aware of mood changes and measures to be taken if needed.

Recommendations

I) Care givers should be counselled to prepare the children for pubertal changes.
ii) Menstrual hygiene should be taught to the children at the appropriate time.


Children with DS have a characteristic behavioral phenotype where language is the most impaired domain of functioning. This forms the greatest barrier to independent meaningful inclusion in the society. Children with Mosaic DS are noted to have better outcomes for language and cognitive abilities.

The DS phenotype is usually characterized by relative strengths in nonverbal communication skills like imitation and gestures, while verbal processing is slow compared to typical developing children of same mental age. They are often reported to progress; though delayed, through stages and sequences of prelinguistic early communication and later language development in a pattern similar to typically developing children. The general profile of language difficulties in children are in expressive language, compared to language comprehension. These problems arise from the anatomical differences of the facial structure and due to the prevalence of hearing loss from frequent middle ear infections, which has a high incidence in the first year.

Social knowledge is the ability to analyze and reason social situations in relation to social rules necessary for social skill and social behavior development. Studies have shown that social functioning is good, but social understanding is poor compared to their normal peers. Poor vocabulary and low-requesting behavior are important predictors of social problems in these children. It is more useful to look at the sequence of development achieved, rather than the age at which it is reached.

Recommendations

I) Functional language and social assessment to be done from the age of one year onwards.
ii) Instructions should be short and clear. Teach children to hear and imitate, find their capability in sound discrimination.
iii) Concrete visual representation of language concepts to be presented.
iv) Parent-mediated intervention plans are encouraged, supporting an everyday bidirectional interaction in naturalistic settings.
v) Social skills training in understanding social situations is beneficial.

INTERVENTIONAL PROGRAMS, EDUCATIONAL AND VOCATIONAL ASPECTS



Physical therapy with speech stimulation is to be started at birth or at least by 3 months of age [24]. Developmental therapy, occupational therapy, behavior therapy and cognitive therapy along with physical therapy and speech therapy should be initiated at the appropriate time. Psychological evaluation and support should be given as the child grows older. Management of attention deficit hyperactivity disorder (ADHD) (49%) and autistic disorders (16%) can manifest as early as 2 or 3 years of age and they should be addressed at the appropriate time. Psychiatric support, especially during adolescent age, is not to be ignored as they can have issues related to sexuality, anxiety and depression, as in any other adolescent. Support of the family, including siblings, should not be neglected, especially in the initial years of life.

Inclusive education is recommended, with special education facility at early school age, as needed. They should be enrolled in the first standard of a normal school by 7-8 years of age, based on their intellectual status. Many of the children have pursued vocational courses and some of them have become graduates. These children are highly trainable, and based on their aptitude; vocational training should be started by about 15 to 17 years (after completion of school). A suitable independent vocation should be encouraged and is preferred to group vocational activities, which should be reserved for children with moderate to severe intellectual challenges.

Recommendations

I) Early interventional programs should be started at least by three months of age.
ii) ADHD and autistic behavior should be looked for, early, and managed.
iii) Psychiatric support should be given for the adolescent with DS and their parents.
iv) Inclusive education in a normal school should be advocated.
v) Vocational training to be initiated based on their aptitude.

It is important to understand and empathize with the parents and find time to respond to all their queries. A proper counselling is mandatory so that they understand the need for a long-term regular follow up with stress on social and rights issues. Age wise follow-up of important medical issues till 18 years are given in Table II and detailed here.

At birth: Karyotyping and genetic counseling, CBC, thyroid function test (TFT), electrocardiogram (ECG), echocardiogram, systemic examination. Record head circumference (HC), weight and length. If antenatal ultrasonogram showed any abnormality, screen postnatally. Eye examination at 4-6 weeks should be done to exclude congenital glaucoma, cataract and other eye abnormalities.

6 months: TFT, full audiological review, record HC, weight and length; plot in DS specific growth charts

1 year: CBC, TFT (Free T4 and TSH once in 3 months, if hypothyroid), dental opinion, hip and foot evaluation in the first year of life. Monitor growth and development.

2 years: CBC, TFT, full ophthalmologic examination, audiology review if not done at 18 months, dental review, testicular examination, monitor growth and development.

3 years: CBC, TFT, audiology review, dental review, testicular examination, growth and development,

4 years: CBC, TFT, ophthalmology, audiology and dental review, testicular examination, growth and development,

5 years: CBC, TFT, audiology and dental review, testicular examination, growth and development.

6-9 years: TFT yearly, 2 yearly audiology and ophthalmology review, annual dental examination, testicular examination, growth and development. Life skills programs with special focus on inter-personal relationships to be initiated.

10-18 years: Continue as in 6-9 years. In puberty: discuss physical and psychological changes, need for gynecologic care in pubescent females. Interventional programs: Developmental therapy, physical therapy, occupational therapy, speech stimulation/ therapy, behavior therapy, cognitive therapy, psychological support for child and family. Early interventional programs should be started at least by 3 months of age.

CONCLUSIONS

Persons with DS have only mild to moderate intellectual disability and have high potential to develop skills in various fields. Early interventional programmes along with early detection and management of associated systemic issues can help these persons lead a disease free independent life. The care-givers should be counselled about the condition and the need for sustained interventional therapy as advised; the medical personnel should be sensitized and trained to provide the right management using a uniform country specific protocol.

Table II Follow-up of Important Medical Issues in Children With Down Syndrome Till 18 Years of Age

	At birth	6 months	1 yr.	2 yrs.	3 yrs.	4 yrs.	5 yrs.	6-9 yrs.	10-18 yrs.
Karyotyping and genetic counselling	√	-	-	-	-	-	-	-	-
Echocardiography	√	-	-	-	-	-	-	-	-
Thyroid function tests	√	√	√	√	√	√	√	√ a	√a
CBC and PBS	√	-	√	√	√	√	√	-	-
Audiology / hearing	√	√	-	√ c	√	√	√	√ b	√ b
Ophthalmology	√	-	-	√	-	√	-	√ b	√b
Dental	-	-	√	√	√	√	√	√ a	√ a
Testicular examination	-	-	-	√	√	√	√	√ a	√ a
Growth / development	√	√	√	√	√	√	√	√ a	√ a

√: indicates evaluation to be done; - indicates evaluation not to be done, unless clinically indicated. aEvery year; bonce in 2 years; cat 2 years if not done at 18 months. Routine vaccinations should not be skipped; and the need for pneumococcal conjugate/ polysaccharide vaccine, influenza and varicella vaccines should be stressed. Evaluation and investigations to be done more frequently, if warranted.

Our thanks to Dr. Shaji Thomas John and other specialists of IAP for this detailed document that will be of immense help to our parents.

Source: Consensus Statement of the Neurodevelopmental Pediatrics Chapter of Indian Academy of Pediatrics (IAP) on the Management of Children With Down Syndrome

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