

Caring for children with Down syndrome: a medical checklist

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Down syndrome (DS) consists of the largest group of children with mental retardation due to a single recognizable syndrome. Once the diagnosis of DS is made (by the paediatrician, neonatologist or family physician), it ushers in a series of investigations for associated medical problems. This is because in this syndrome almost every system or organ in the body needs special attention. Appropriate interventions, if carried out on time, can reduce complications and improve the quality of life of these children. It is therefore important that all doctors caring for children with Down syndrome are updated with the latest recommendations and are mindful that they should avoid inflicting undue hardships on the family.

This article presents an age specific preventive medical checklist for use by paediatricians, family physicians and others. Its objective is to improve the health of DS children. The educational aspects have not been included in these guidelines although the care of DS involves families and educators in addition to health professionals.

Usually protocols and medical guidelines are the result of deliberations of committees and experts. I have put together here accepted current practices based on standards stipulated by several recognized medical organizations. They reflect recent advances acceptance by the Down Syndrome Medical Interest Group (UK) and American Academy of Paediatrics in USA. Suitable modifications for local adaptation have been made taking into account availability of expertise and referral pathways. It is hoped that these checklists will constitute a feasible programme of medical care for children and adolescents with DS.

Birth - one month

- A complete neonatal examination.
- Break news to parents.
- Address parental concerns.

- Careful cardiac evaluation including electrocardiograph (ECG).
(ECG is a useful screening tool even if there are no abnormal signs)
- Refer to a paediatric cardiologist for echocardiogram if cardiac defect is suspected.
- Check haemoglobin & haematocrit for polycythaemia.
- Test thyroid function with serum TSH estimation.
(Skeletal age is not a suitable alternative as DS itself can retard skeletal maturation)
- Exclude duodenal atresia and other gastrointestinal abnormalities (if indicated).
(If vomiting, duodenal atresia or duodenal stenosis MUST be excluded.)
- Enroll help of lactation nurse or midwife if feeding difficulties are present.
- Early ophthalmological referral if absent red reflex, nystagmus or strabismus.
- Offer family support by introduction to support / parent group.
- Down syndrome should be written on Child Health Development Record (CHDR) as "Reason for special care".

One month – 12 months

- Cardiology referral is essential by three months (if not carried out earlier).
(This should take place even in the absence of a murmur)
- Monitor for progressive pulmonary hypertension (even in absence of heart failure).
(Early onset of pulmonary hypertension is a feature in DS)
- Explain benefits of early cardiac surgery*.
- Issue special DS growth charts (for weight and height).
- Serially chart weight and height on both DS & normal growth charts.
- Routine immunization (same as for well child).

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- Hearing assessment at 3 months with auto acoustic test (OAE) or auditory brainstem response (ABR).
- Repeat hearing assessment in 6 months.
- Ophthalmological evaluation at 6 months (earlier if concerns of poor vision).
- Additional TSH screening at 6 months if neonatal screening was normal and hypotonia is prominent.
(Worsening of hypotonia may be an indication of heart failure)
- Developmental assessment and therapy.
(Refer to a development clinic if available locally)
- Explain need for home based early stimulation programmes.
- Refer for occupational, speech and physiotherapy.
- Visualize tympanic membrane with every acute respiratory tract infection.
- Investigate for duodenal stenosis if vomiting persists.
- Consider Hirschprung disease if constipation responds poorly to stool softeners.
- Reinforce family support.
- Offer genetic counselling karyotype if indicated.

* *Shunt operations for AV canal defects and Tetralogy of Fallot are best performed before the age of two years.*

One year – five years

- Reassess development periodically, monitor compliance and document progress.
- Recommend group therapy where possible.
- Calculate ‘weight for height’ every six months.
(Failure to thrive requires the usual investigation workup)
- Six monthly hearing evaluations, if abnormality suspected, until 3 years of age.
- Eye evaluations at 2 years and school entry.
- Twice daily brushing of teeth should be recommended.
- Dental examination at 2 years and as appropriate thereafter.
- Estimate TSH level annually.
- Subacute bacterial endocarditis (SABE) prophylaxis prior to minor surgery or dental procedure if cardiac lesion present.
- Pneumococcal vaccine if severe cardiac or respiratory disease.
- If persistent vomiting is a problem duodenal stenosis needs investigation.

- Chronic constipation not responding to stool softeners needs exclusion of Hirschprung disease.
- Severe recurrent chest infections may need immunological investigations.
- Pallor may require evaluation for leukaemia
- Diagnosis of autism needs careful evaluation.
(Repetitive movements such as flapping of hands and speech delay may be due to severe retardation)
- Attention deficit hyperactivity disorder (ADHD) should be a diagnosis of exclusion.
(Poor attention and restlessness may be due to poor vision, impaired hearing or learning difficulties and ADHD should be a diagnosis of exclusion only).
- At 4 years, in preparation for school, a detailed speech evaluation is necessary.
- Speech therapist should provide a programme to maximize language development with augmentative hearing device if necessary.
- Refer for financial support from social services department.
- Guide to appropriate schooling and individualized education programme (special education or mainstream).

5 years – 12 years

- Annual educational and family needs assessment.
(A community paediatrician would be an appropriate professional)
- Monitor “weight for height”.
(If obese give appropriate dietary advice, check liver function)
- Give dietary advice, (a balanced high fibre diet) with a caloric intake lower than the recommended daily allowance (RDA) for same height and weight (provided there is no growth failure).
- Recommend regular exercise and recreational programmes.
- Monitor for behaviour problems and refer to mental health services if necessary.
- Annual TSH & T4 estimations.
- Annual hearing and vision assessments are important especially during schooling.
- Annual dental checks.
- Monitor for otitis media at each episode of acute upper respiratory tract infection.

- Inquire for sleep problems and treat accordingly (*Snoring, mouth breathing, restlessness, excessive daytime sleepiness, unusual sleep position, difficulty in focusing and learning new information are suggestive clinical features*)
- SABE prophylaxis and cardiac follow up as appropriate.
- Radiographic screening for atlanto-axial instability to be carried out if any suspicion of this complication or if required for sports. (*X rays prior to selected sports - gymnastics, high jump, diving, football - are no longer mandatory*)
- Behavioural problems should be addressed

12 years – 18 years

- Monitor for obesity.
- SABE prophylaxis if indicated.
- Routine screening with echocardiogram at 18 years for mitral valve prolapse (*Even in the absence of congenital heart disease acquired heart disease may occur*)
- If neurological examination suggests spinal cord compression refer for radiological assessment by measuring atlanto-dens distance (lateral neck X-rays in neutral, flexion & extension positions) (*Gait abnormalities, incontinence, tendon & plantar reflexes are warning signs*)
- Encourage socializing – friends, scouting /guiding, & extra curricular activities such as music, sports, drama and dance.
- Psycho-educational evaluation by child psychiatrist every 2 years.
- Ensure life skills (e.g. money-handling skills, dressing, cooking) are included in educational programme.
- Discuss long term plans e.g. living arrangements, sheltered employment.
- Encourage to follow vocational training programme.
- Address sexual issues, if necessary.

N.B. Specific medical investigations and complications are not elaborated at great length in this article and further reading is recommended if necessary.

In recent years, many treatment modalities have claimed to “cure” Down syndrome. No account of ‘current practice’ would be complete without these. This information is on the internet and with

increasing numbers of parents accessing this source, it is important that those caring for Down syndrome are aware of the truth about these so called “alternative” therapies.

Unconventional therapies

High dose vitamins, supplemental zinc, supplemental selenium, freeze dried fetal cell injections and piracetam (a medication classified as a cerebral stimulant) are among these. Scientific groups interested in Down syndrome have extensively studied these manifold therapies. Their conclusions are that these treatment methods do not improve intellect and have in some cases even proved harmful. As regards piracetam a double blind placebo controlled study on twenty Down syndrome children treated with it found the medication ineffective.

Facial plastic surgery to alter characteristic facial features is another such recommendation. It claims greater social acceptance. This procedure is controversial because faces continue to grow into adolescence. It is therefore not a medically indicated procedure on children and many insurance companies do not pay for this procedure. Neither is tongue reduction surgery recommended because it has not been shown to improve expressive speech.

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