Medical management of children with Down syndrome

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Abstract

Down syndrome, trisomy 21, is the most common autosomal trisomy, and commonest identifiable cause of learning disability. Despite current prenatal screening practice birth prevalence continues to be around 1/1000 live births.

Children with Down syndrome have an increased risk of congenital abnormalities and a wide range of treatable medical problems. Paediatricians have a key role in ensuring that these are recognized and treated so that the children's progress is not hampered by additional secondary but preventable disability, and health problems do not prevent them reaching their potential.

In this article we consider the Paediatrician's role with a suggested approach to medical management throughout childhood, and a review of the most frequently occurring health issues. These include cardiac, respiratory, gastrointestinal and haematological disorders, thyroid dysfunction, hearing and vision problems, cervical spine and other orthopaedic problems, immune and autistic spectrum disorders, growth and sexual development.

Keywords Down syndrome; guidelines; trisomy 21

Down syndrome is a common cause of developmental disability. There is widespread awareness of the associated physical features and variable learning disability, but possibly less understanding of the wide range of health problems which may also affect those with the syndrome.

Increasingly diagnosis of Down syndrome is made antenatally and many affected pregnancies are terminated. Data from the National Down Syndrome Cytogenetic Register show that in England and Wales 90% of women with antenatal diagnosis of Down syndrome choose to terminate. Despite this there has been no major change in birth prevalence, probably because women are now starting their families later, and as incidence of Down syndrome rises with maternal age there are likely to be more conceptions of babies with Down syndrome. Babies born to those who decide to continue the pregnancy after diagnosis, together with those diagnosed after birth (false negative, or screening not performed), currently give a live birth rate of 1.0/1000 in the UK. It is therefore likely that there will continue to be more than 700 babies with Down syndrome born each year in England and Wales.

Paediatricians have a key role in health provision for these children. Whilst some thrive from an early age and are in good health throughout childhood there is, among the group as a whole, an increased risk of congenital abnormalities and a wide range of medical problems (Box 1). The impact of these problems on general health, growth and development may be even greater than would be expected for other children because of the associated developmental delay and learning disability.

Historically, some treatable conditions were thought to be 'part of the syndrome' and left untreated. There may have been lack of recognition of potential benefits to overall functioning of the child or even discrimination. Today we hope that the health of children with Down syndrome will be monitored as carefully as that of any child, and treatment offered when necessary so that their progress is not hampered by additional secondary but preventable disability, and health problems do not prevent them reaching their potential.

In this article therefore we consider the Paediatrician's role in ensuring that this is achieved. Inevitably our review cannot be comprehensive but we cover all major and some less well known health problems, and less common problems are merely listed in Box 1.

The Paediatrician's role

At time of diagnosis

Diagnosis of Down syndrome is increasingly made antenatally. The Paediatrician may be called on to give information to parents in order to help them reach a decision about the future of the pregnancy. However for many babies diagnosis is suspected at or soon after birth when physical features are noticed. It is essential that an experienced Paediatrician is then involved immediately in

- Confirming the diagnosis
- Giving information to the parents
- Medical assessment

Confirmation of diagnosis by chromosomal analysis is usually possible within 48 hours. Most parents prefer to be told of the suspected diagnosis as soon as possible, and disclosure should not be delayed until chromosome confirmation. The importance of disclosure of diagnosis and the way it is handled cannot be over-stressed. Parents often recall many years later how they were told and the information given. These initial discussions will influence how the parents adjust to the diagnosis and how they view their baby. The news must be shared sensitively, honestly and non-judgmentally. Information given in these early days must be up-to-date, balanced, and include positive aspects as well as describing the difficulties that may be faced. It should include

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Some specific medical problems that occur more frequently in people with Down syndrome

Cardiac
Congenital malformations
Cor pulmonale
Acquired valvular dysfunction

Orthopaedic
Cervical spine disorders
Hip subluxation/dislocation
Patellar instability
Scoliosis
Metatarsus varus
Pes planus

ENT and Respiratory
Respiratory infections
Conductive hearing loss
Sensorineural hearing loss
Sleep related breathing disorders
Chronic catarrh

Ophthalmic
Refractive errors
Blepharitis
Nasolacrimal obstruction
Cataracts
Glaucoma
Nystagmus
Squint
Keratoconus

Gastrointestinal
Congenital malformations
Feeding difficulties
Gastro-oesophageal reflux
Hirschprung's disease
Coeliac disease

Endocrine
Hypothyroidism
Hyperthyroidism
Diabetes

Immunological
Immune dysfunction
Autoimmune diseases, e.g. inflammatory arthropathy, diabetes, thyroid disorders, alopecia

Haematological
Transient neonatal myeloproliferative states
Leukaemia
Neonatal polycythaemia
Neonatal thrombocytopenia
Polycythaemia, macrocytosis, leucopenia

Dermatological
Dry skin
Folliculitis
Vitiligo
Alopecia

Neuropsychiatric
Infantile spasms and other myoclonic epilepsies
Autistic spectrum disorder
Depressive illness
Dementia (adults)

Box 1

- Aetiology of Down syndrome (why did it happen? will it happen again?)
- Likely developmental progress (discuss wide variation in abilities)
- Immediate and if appropriate possible future health concerns
- Local resources
- Long-term prognosis (life expectancy, what the future may hold)

Parents should be given contact information for the local Down's syndrome association (Box 2). Written information should be given and the Early Support Programme materials are useful, as well as the Personal Child Health Record (PCHR) insert[b] for babies born with Down syndrome. Some parents appreciate being put in touch with another family with a child with Down syndrome. They should leave hospital knowing when they will next be seen, and who they can contact in the meantime should the need arise. The primary healthcare team should also be informed.

The neonatal period

Every newborn with Down syndrome should have a comprehensive medical assessment focussing on medical problems associated with the syndrome. Malformations of the gastrointestinal tract usually present clinically but other problems may not. Even major congenital heart disease may not be evident on initial clinical examination and appropriate investigative screening and expert cardiac assessment must be initiated before discharge from hospital. The eyes should be checked for cataract. Universal newborn hearing screening and blood spot TSH screening are particularly important, given the increased prevalence of hearing problems and hypothyroidism in Down syndrome. Recent research also supports checking a neonatal full blood count (FBC) for transient myeloproliferative disorder.

Some newborns with the syndrome require a significant period of special care. Average birth weight is 3.07 kg and time to regain birth weight may be a month or more. Early feeding difficulties are common but with the right support breast feeding can be achieved by most.

Ongoing medical management

Children with Down syndrome should be offered regular medical review by a Paediatrician throughout childhood. This may be via a hospital department, child development centre, or community paediatric service. The type of service offered will