Ear, nose and throat problems in children with Down syndrome

Down syndrome (trisomy 21) is a common childhood condition. As many of the features involve the head and neck, the diagnosis of Down syndrome has significant implications for the ear, nose and throat surgeon.

A 'syndrome' is a collection of features which occur together and go to make up a characteristic clinical entity. John Landon Down described the commonest of these syndromes as 'Mongolian idiocy' in 1866. In 1959, Lejeune discovered the chromosomal abnormality – trisomy 21.

Prevalence

Down syndrome occurs in 1 in 700 live births. About 50% of pregnancies where the fetus has trisomy 21 end in spontaneous abortion. There is an association with increased maternal age. The condition can be diagnosed with a high degree of sensitivity and specificity in the prenatal period. Commonly used tests are chorionic villus sampling and amniocentesis. Improvements in prenatal sonography and maternal serum testing permit accurate screening with a low morbidity (Alfirevic and Neilson, 2004; Nicolaides, 2004).

General features

The characteristic features are a typical dysmorphic facies with flattening of the nasal bridge, epicanthal folds, anteroposterior flattening of the skull, learning disability, hypotonia and orocranial anomalies.

The head and neck

Many of the features of Down syndrome involve the head and neck (*Figure 1*). Down's children will usually have small ears and a typical Down's facies characterized by hypoplasia of the midface. The palpebral fissures are upslanting. The baby may be hypotonic or floppy. Often the lips are large although they may be of normal size at birth. The diagnosis can be difficult in the newborn as the characteristic facies only develops in later childhood. The child may have a high arched palate, not infrequently with a cleft. There is micrognathia and a tendency towards dental and periodontal problems.

Associated disorders

Some 40% of Down's children have associated heart disease. This can vary from mild disorders such as ventricular septal defects which close spontaneously to major congenital cyanotic heart disease which may be fatal. About 15% have major anomalies of the gastrointestinal tract. These include duodenal atresia and tracheo-oesophageal fistula. There is a tendency towards CNS disease and Down's children may develop a form of Alzheimer's disease in adolescence. They may have haematological abnormalities such as a tendency to develop leukaemia. Many Down's children will have a learning disability. The IQ range is usually from 40–90. About one tenth of the population of children with learning disability will have Down's syndrome.

Ear, nose and throat problems

Ear, nose and throat (ENT) problems (*Table 1*) include upper respiratory tract infections, rhinitis, otitis, airway obstruction, deafness, thyroid disorders and sialorrhoea or drooling (Kanamori et al, 2000; Venail et al, 2004).

Airway disorders

The Down's child will often have problems with the airway (*Table 2*). Typically, the calibre of the subglottis and trachea is small. The pharyngeal airway can have an unusual shape. The child may have tracheomalacia or

Figure 1. Girl (aged 4 years) with Down syndrome.

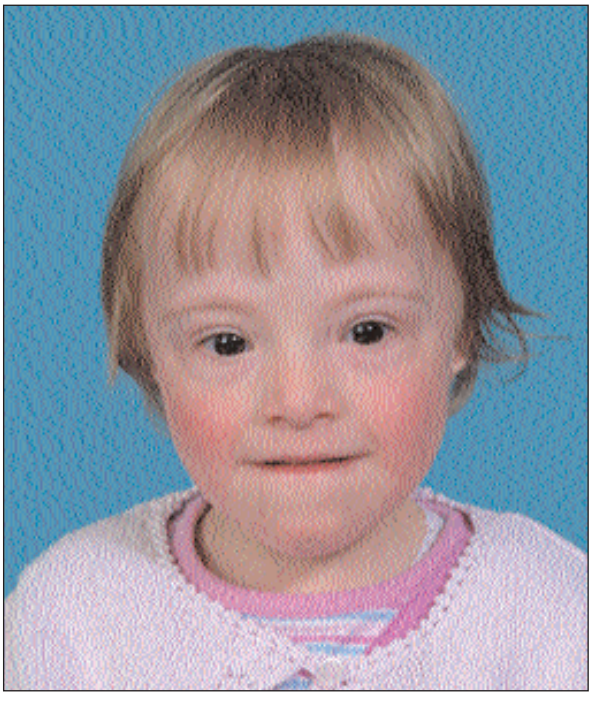

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Table 1. Ear, nose and throat problems in patients with Down syndrome

Frequent upper respiratory tract infections
Airway obstruction
Deafness
Thyroid disease
Sialorrhoea
Atlanto-axial instability (manipulate the head with extreme care under anaesthesia)

laryngomalacia. Macroglossia can be another feature of Down syndrome giving rise to airway obstruction (*Figure 2*). Sometimes children will have tracheo-oesophageal fistula or major abnormalities of tracheo-oesophageal development including laryngotracheal clefts. These can be fatal in early infancy if severe. There is a higher incidence of subglottic stenosis and laryngeal webs.

Foreign bodies

Ingesting foreign bodies is a concern in all children, but is more common in children with a learning disability and in children with hypotonia, neurological dysfunction or abnormalities of swallowing. If a Down's child presents with drooling and dysphagia it is important to bear in mind that he/she may have bolus obstruction of the oesophagus (*Figure 3*). If a Down's child presents with unexplained recurrent chest infections it is prudent to consider foreign body aspiration with obstruction of the bronchus. A chest X-ray can be helpful (*Figure 4*).

Figure 2. Macroglossia – an important component of airway obstruction.

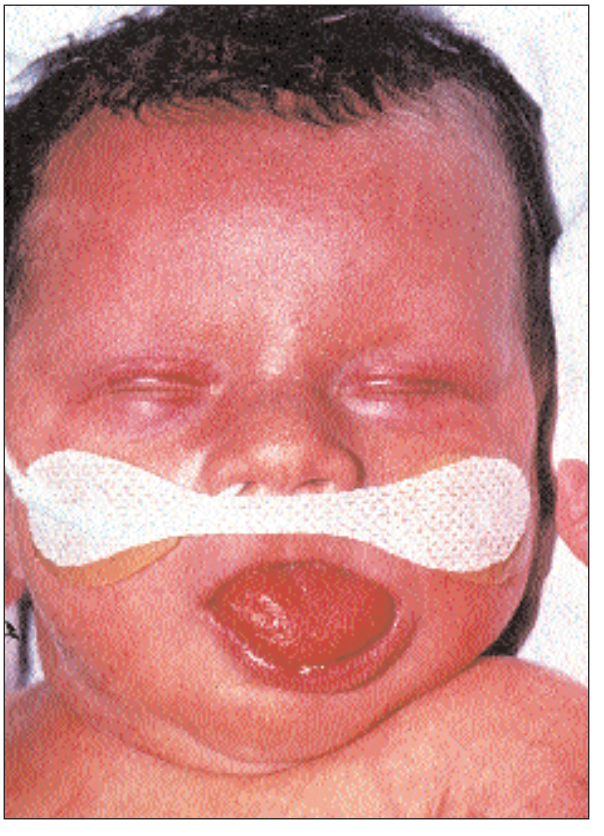


Table 2. The airway in Down syndrome

Narrow subglottis and trachea – may need smaller endotracheal tube during anaesthesia
Midfacial hypoplasia – increased incidence of obstructive sleep apnoea
Hypotonic pharyngeal airway
Macroglossia

Obstructive sleep apnoea

Up to 50% of Down's children will have obstructive sleep apnoea. A number of factors will bring this about; one is the facial profile with hypoplasia of the midface.

Figure 3. Ingested toy causing oesophageal obstruction.

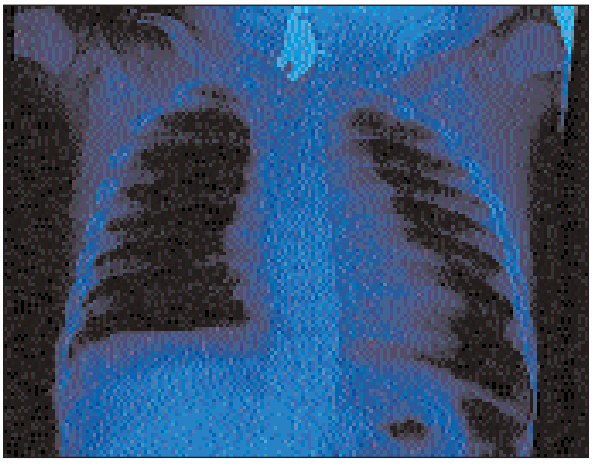
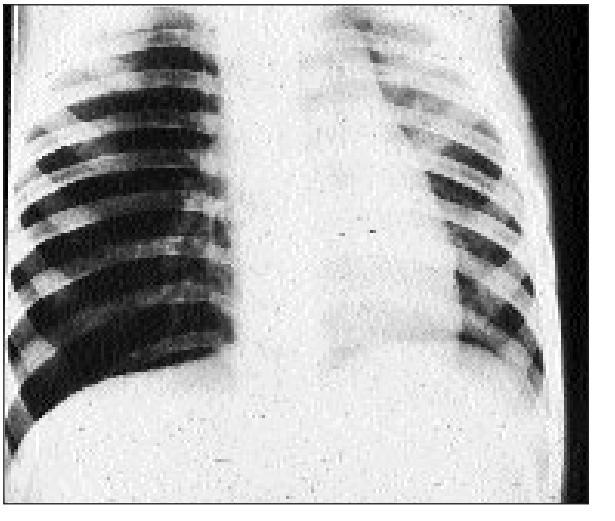


Figure 4. Foreign body in the right bronchus. Chest X-ray shows hyperinflation of the right lung as a result of partial obstruction. A piece of orange peel was recovered at bronchoscopy.



Macroglossia, pharyngeal abnormalities, hypotonia and central factors also contribute. Adenoidectomy is often unhelpful but early tonsillectomy may bring about considerable improvement.

Anaesthesia in Down syndrome

Children with Down syndrome need to be looked after by an experienced anaesthetist. They can be difficult to intubate, difficult to extubate and are often slow to recover. Some will have atlanto-axial instability. Often a Down's child will not be able to take an age-appropriate endotracheal tube, but need a size smaller.

Atlanto-axial instability

This mainly occurs in children up to the age of 10 years. It is more common in females. It is thought to be caused by laxity of the cervical ligaments. Manipulation of the neck in Down's children during surgery and anaesthesia should be undertaken with extra care. Catastrophic neurological sequelae have been described resulting from trauma to the spinal cord.

Deafness and hearing problems in Down syndrome

Down's children have a small pinna, and the ear canal is narrow. They have a tendency to produce a lot of ear wax. They may have ossification of the cochlea and 70% develop deafness. It is vitally important that Down's children are screened for deafness at birth (*Table 3*). Some will have ossicular fixation, persistent mesenchymal tissue in the middle ear, or the Mondini malformation – a major structural abnormality of the cochlea causing profound hearing loss. Deafness can be conductive, sensori-neural or mixed. Rehabilitation of hearing can bring about dramatic improvements in the quality of life of both child and family. It is important to consider conventional or in some cases bone-anchored hearing aids. Some children may be suitable for cochlear implantation but all need regular and skilled audiological surveillance.

Table 3. Ear disorders in Down syndrome

Frequent acute otitis media and otitis media with effusion
Small pinna and external meatus
Premature ossification of the cochlea
Facial nerve dehiscence common

Otitis media

Otitis media is more common in Down's children (*Figure 5*). Their non-specific defence mechanisms are poor, they have abnormalities of the Eustachian tube and they may have hypotonia which interferes with normal Eustachian tube closure. 'Glue ear' or otitis media with effusion is very common. Grommets can be troublesome and may be associated with persistent perforations. Some Down's children are better managed with early fitting of hearing aids.

Mastoid surgery is particularly challenging in Down's children. They may have dehiscence of the facial nerve and a small external auditory meatus.

Conclusions

Children with complex needs – particularly exemplified by Down syndrome – are often best looked after in a paediatric centre. These children have multiple complex needs and require multidisciplinary input to optimize care. BJHM

Conflict of interest: none.

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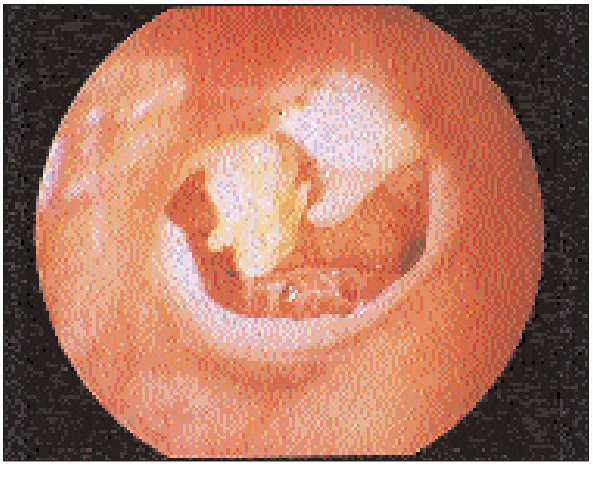
Figure 5. Perforated ear drum in long-standing otitis media.



KEY POINTS

- Down syndrome is the commonest known chromosomal abnormality.
- Many of the features involve the head and neck.
- Airway obstruction, otitis media and deafness are particularly prevalent in Down's children.
- Multidisciplinary care can bring about a dramatic improvement in quality of life for these children and their families.
- Anaesthesia and surgery for Down's children is best carried out by experienced clinicians with good support facilities.