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MAIN ARTICLE

Current demand of paediatric otolaryngology input for children with Down's syndrome in a tertiary referral centre

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Abstract

Objective: This study aimed to evaluate the activity of paediatric otolaryngology services required for children with Down's syndrome in a tertiary referral centre.

Methods: A review of the paediatric otolaryngology input for children with Down's syndrome was performed; data were obtained from the coding department for a two-year period and compared with other surgical specialties.

Results: Between June 2011 and May 2013, 106 otolaryngology procedures were performed on children with Down's syndrome. This compared to 87 cardiac and 81 general paediatrics cases. The most common pathologies in children with Down's syndrome were obstructive sleep apnoea, otitis media, hearing loss and cardiac disease. The most common otolaryngology procedures performed were adenoidectomy, tonsillectomy, grommet insertion and bone-anchored hearing aid implant surgery.

Conclusion: ENT manifestations of Down's syndrome are common. Greater provisions need to be made to streamline the otolaryngology services for children and improve transition of care to adult services.

Key words: Down Syndrome; Otolaryngology; Obstructive Sleep Apnea; Tonsillectomy; Otitis Media; Otorhinolaryngologic Surgical Procedures

Introduction

Trisomy 21 is the most common chromosomal abnormality, affecting 1 in 600–800 live births.[1,2] There are over 40 000 people in the UK with Down's syndrome.[3] Advances in the management of the cardiac abnormalities during the neonatal period of Down's syndrome babies has led to a significant improvement in their life expectancy,[1] to approximately 60 years of age.[2–4] It is the management of these cardiac abnormalities that is the dominant focus during the neonatal period. However, otorhinolaryngological manifestations of Down's syndrome are well documented, and form a major requirement of care for these individuals and their families.

Airway problems affecting children with Down's syndrome include large adenoids and tonsils combined with macroglossia. Sleep-disordered breathing is the most common respiratory disorder, with a prevalence ranging from 31 to 79 per cent.[1,2,5] These children have a number of structural and dynamic features that predispose them to obstructive sleep apnoea (OSA), including small midface size, narrow nasal passages, hypotonia of the oropharyngeal and hypopharyngeal muscles, macroglossia, and adenotonsillar hypertrophy.[1] These features also predispose children with Down's syndrome to post-operative complications, such as apnoeas, hypoxaemia and pulmonary oedema.[1]

Current recommendations for paediatric patients with Down's syndrome suggest discussion of OSA symptoms with parents by the age of six months, and assessment by the age of four years.[6] A study investigating OSA in young infants with Down's syndrome found that the average female age at diagnosis was 58 days and the average male age was 20 days.[7] That study also found a correlation between OSA severity and younger age. Furthermore, OSA seemed to coexist more frequently in patients with dysphagia and gastroesophageal reflux disease.[7]

The otological manifestations associated with Down's syndrome include a small pinna, with narrow or even stenosed external ear canals, and an increased incidence of glue ear. Affected patients can have abnormal ossicles and suffer with a mixed hearing loss. The sensorineural aspect of hearing loss tends to be progressive and in the high frequency range.[1] There are several options for the management of hearing impairment, including hearing aids, implantable hearing devices and grommets. Thus, significant input is required from

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audiological services, speech therapists and teachers of the deaf. These difficulties can continue from childhood into adulthood, and it is therefore essential that these individuals are not lost in the system at the transition point.

About 15 per cent of patients with Down's syndrome are at risk of atlanto-axial joint subluxation.[8] Hence, it is essential that these individuals are managed at all times by senior, experienced clinicians who are aware of such potential issues. Subglottic stenosis, laryngotracheomalacia and vocal fold palsy may also be present. The multisystem involvement of Down's syndrome requires a multidisciplinary approach. Moreover, with an increasing life expectancy, care needs to be continued from childhood to adulthood, with an emphasis on primary care involvement.

The Down's Syndrome Association has an extremely useful website, providing support and crucial information for patients with Down's syndrome and their carers. It also gives health professionals key information to understand and maximise care for patients with Down's syndrome.[3]

This study aimed to evaluate the activity of ENT services required for children with Down's syndrome in a children's tertiary care centre. The results were compared with other surgical specialties, with an aim of making recommendations to streamline care for children with Down's syndrome.

Materials and methods

Data were retrospectively collected from June 2011 to May 2013 (a 24-month period). The data collected, obtained from the coding department at Birmingham Children's Hospital, UK, included: the ENT procedures performed on all Trisomy 21 patients for all surgical specialties, the co-morbidities of these children and the length of their hospital stay.

Results

There are seven paediatric otolaryngological consultants at the tertiary care centre. A dedicated clinic is held once per month for children with Down's syndrome; this clinic is multidisciplinary as it utilises the services of an audiologist and a speech therapist.

Operating theatre staff at Birmingham Children's Hospital are very familiar with Down's syndrome and the careful handling that is required, in particular when positioning the patient for intubation. All intubations are performed with the patient in the neutral position, and gentle head rotation is the standard.

The average age of the children who underwent surgical treatment within ENT, during the study period, was 6.1 years, with an age range of 6 days to 17 years. Most patients were managed as day cases; about one-fifth were hospitalised for 1 night and one-tenth required more than 1 night's admission.

Table I shows that ENT procedures were the most frequently performed procedures in these children with Down's syndrome. This was followed by cardiac surgery and general paediatric surgery. The type of ENT surgery most commonly performed was adenoidectomy, followed by grommet insertion, tonsillectomy and bone-anchored hearing aid implant surgery, as illustrated in Figure 1. Table II shows that the most common pathologies encountered in children with Down's syndrome were obstructive sleep apnoea, otitis media, hearing loss and congenital heart disease.

TABLE I
PROCEDURES PERFORMED ON DOWN'S SYNDROME PATIENTS IN EACH SPECIALTY*

Surgical specialty	Number of procedures
ENT	106
Cardiology	87
General paediatrics	81
Trauma & orthopaedics	14
Neurosurgery	9
Urology	7
Maxillofacial	4
Plastics	4
Ophthalmology	4

*Between June 2011 and May 2013

Discussion

A survey of patients attending a Down's syndrome conference revealed that 50 per cent consulted an otolaryngologist regularly.[9] Given the complex and multifactorial nature of their head and neck manifestations, a multi-professional approach is required, with the involvement of audiologists, speech and language therapists, general practitioners, and paediatricians. As the life expectancy of patients with Down's syndrome is increasing, more aggressive and proactive care should be delivered to these individuals. Increased survival is not only associated with a longer period of care but is also related to a longer period of more specialised needs.

Our results showed that ENT procedures were the most common procedures carried out in children with Down's syndrome, followed by cardiac surgery and then general paediatric surgery. This confirms that the ENT manifestations are a dominant focus of morbidity in children with Down's syndrome. This condition has a spectrum of clinical pathology, and in some patients multiple coexisting conditions can be found. There should be close links between the paediatric specialties so that care for children with Down's syndrome is coherent and transparent, in particular between the otolaryngologist, the cardiac surgeon, general surgeon and paediatricians. Given the risk of subglottis stenosis and post-operative complications in this group of patients, it may be wiser to perform some procedures under the same general anaesthesia.

The most common ENT procedures carried out on children with Down's syndrome in this study were adenoidectomy, tonsillectomy, grommet insertion and bone-anchored hearing aid implant surgery. Obstructive sleep apnoea was the most common

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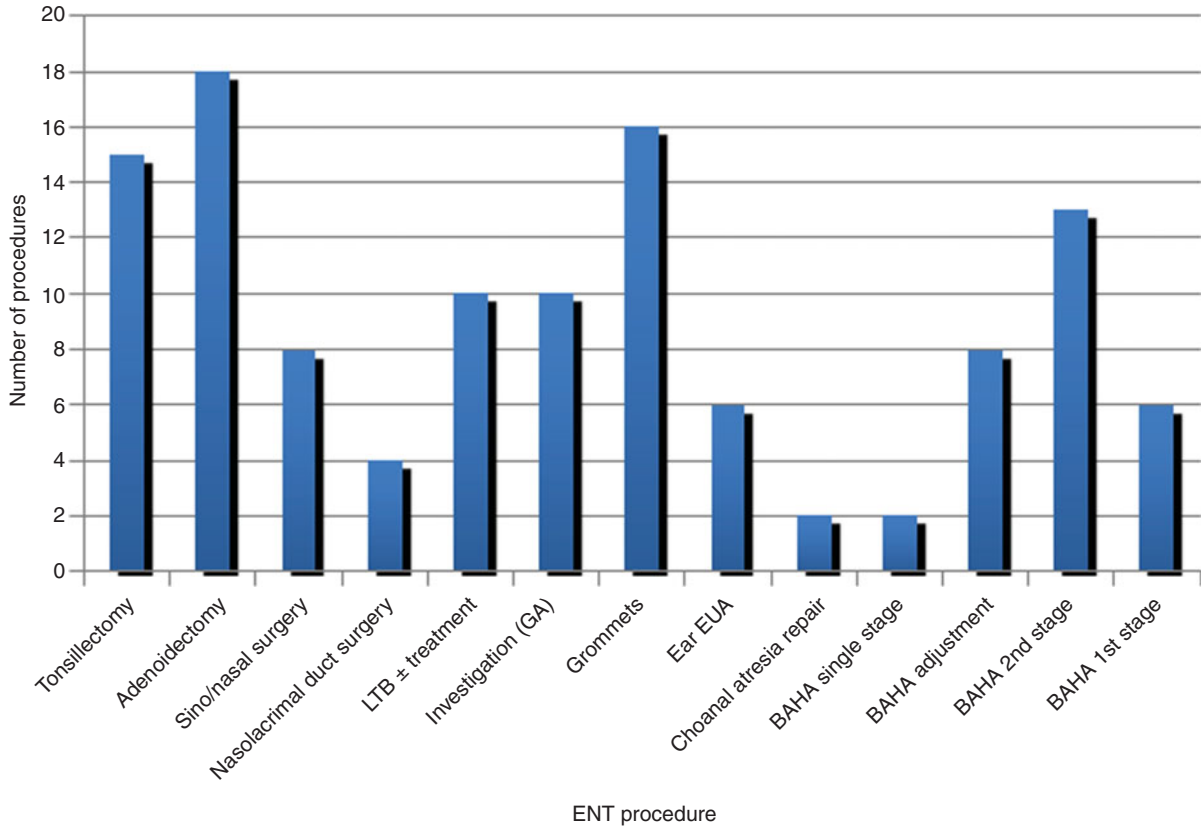

FIG. 1

Graph showing the ENT procedures performed on children with Down's syndrome. LTB = laryngo-tracheo-bronchoscopy; GA = general anaesthesia; EUA = examination under anaesthesia; BAHA = bone-anchored hearing aid

diagnosis in these children, suggesting that adenotonsillectomy was most commonly performed for this pathology.

Marcus *et al*. found that 77 per cent of the patients in their study had abnormal polysomnograms, of which 45 per cent met the criteria for OSA.[10] A focused history, in which patients are asked specifically about 'choking episodes' and witnessed apnoeic episodes, could help in screening children with Down's syndrome for OSA.[1] Another study compared OSA severity in: children with Down's syndrome, a cohort of normally developing children and children with other co-morbidities of comparable severity to Down's syndrome.[5] The researchers found that children with Down's syndrome had worse gas exchange, as reflected in a worse McGill oximetry score, and higher average partial pressure of carbon dioxide during sleep. The children with Down's syndrome were also prone to more severe OSA compared to normally developing children.[5] Nocturnal hypoxaemia has been related to declining functioning in areas of visual spatial ability, mental flexibility and processing speed in adults, and increasing hypoxia is related to increasing OSA severity; this may have significant implications for children with Down's syndrome.[5] Many OSA sequelae, such as pulmonary hypertension and failure to thrive, can be seen in children with Down's syndrome. It is therefore essential that OSA is considered and the necessary investigations are conducted to rule this out early on.

Bull and the Committee on Genetics published a clinical report on the health supervision for children with Down's syndrome for the American Academy of Pediatrics.[6] They recommend that parents of babies one month old and older are advised about OSA symptoms, including heavy breathing, snoring,

TABLE II
COMMON ENT PATHOLOGIES IN DOWN'S SYNDROME CHILDREN REQUIRING SURGICAL INTERVENTION

Anatomy affected	Pathology	*n*
Airway	Obstructive sleep apnoea	29
	Adenoidal or tonsil hypertrophy	8
	Choanal atresia	7
	Laryngomalacia	4
	Subglottic stenosis	1
	Laryngeal web	1
	Other congenital malformation of larynx	6
Ears	Otitis media	27
	Conductive hearing loss	21
	Hearing loss (unspecified)	24
	Otorrhoea	1
Heart	Congenital heart disease	22

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unusual sleep positions, night awakenings with daytime somnolence, apnoeic episodes, and subsequent behavioural issues secondary to disturbed sleep.[6] These babies should then be referred on to a physician who has expertise in paediatric sleep disorders. The report confirms a poor correlation between parental reporting of abnormal sleep patterns and polysomnogram reports, and therefore advises that all children have a sleep study or polysomnogram by the age of four years.[6] Parents should also be advised that obesity is a risk factor for OSA.[6]

With the life expectancy of this group increasing, there is a greater need to manage sleep-related problems to reduce morbidity and improve quality of life. Studies have shown that adenotonsillectomy does improve some OSA parameters in Down's syndrome patients, but not as much as in non-Down's syndrome patients. As compliance with continuous positive airway pressure (CPAP) is low amongst adults,[11] it is likely to be lower in children with developmental delay, and therefore any benefit in OSA parameters should be considered.[12] A study investigating the management options for OSA in neonates and infants aged 0–12 months found that gastro-esophageal reflux treatment was the most common, and adenoidectomy was the most common surgical intervention.[13] In the majority of OSA cases, adenotonsillectomy should be considered as a first-choice treatment, but there should be awareness that it may not be sufficient on its own.[6] In children who do not get sufficient benefit, craniofacial and tongue surgery may be appropriate, and should be considered.[6,14] The next option should be CPAP, which has been found to be effective when tolerated.[14] Repeated physiological studies should be performed to assess the benefit of the intervention introduced.[6] In a very small number of patients, these interventions may not be effective, and a tracheostomy may have to be performed as a last resort to avoid ongoing hypoxaemia and pulmonary hypertension.[6] Overall, CPAP or bi-level positive airway pressure use has been associated with the greatest benefit in terms of apnoea–hypopnoea index.[10]

Patients with Down's syndrome have a predisposition to hearing loss, most commonly due to otitis media with effusion. If left untreated, this can affect behaviour and language development, and should therefore be diagnosed and treated early. Soft or hard band bone conductor hearing aids are the devices of choice to treat children with Down's syndrome. Grommets also play a role; however, the risk of otorrhoea following grommet insertion can be significant. Bone-anchored hearing aids have been used successfully in children with hearing loss associated with Down's syndrome.[15,16]

The incidence of sensorineural hearing loss increases with age, with a prevalence of about 20 per cent in adolescence and early adult life.[17] Presbyacusis develops 30–40 years earlier in patients with Down's syndrome when compared with the general population.[18,19]

Other common pathologies seen in children with Down's syndrome in the current study were otitis media and hearing loss. Shott reported that the majority of stenotic ear canals grow with age, and usually by the age of two to three years they are adequate for examination.[20] Some patients will develop chronic suppurative otitis media thereafter and so continued examination is necessary.

Swallowing dysfunction is often seen in patients with Down's syndrome; in particular, it is the oral and pharyngeal phase of swallowing that is disrupted.[21,22] Oropharyngeal hypotonia, oral sensorimotor deficits and cognitive deficits predispose an individual to pharyngeal dysphagia. Video fluoroscopy is the 'gold standard' investigation for assessing swallowing. This should be considered in all patients with signs or symptoms indicating laryngeal penetration and/or aspiration. Phonological development is influenced by hearing, orofacial anomalies and cognition. These are often abnormal in patients with Down's syndrome, resulting in delayed speech or articulation problems.[23] De-institutionalisation of children with Down's syndrome and a push for mainstreaming must be accompanied with additional support in order to optimise the wellbeing of these patients.[23] Therefore, the role of speech therapy and dysphagia professionals in the management of Down's syndrome in children is vital.

There are several anaesthetic considerations that need to be taken into account in patients with Down's syndrome, including airway and respiratory system anomalies and potential cervical spine instability. Bull *et al*. recommended that parents of children with Down's syndrome should be advised from birth onwards regarding the importance of cervical spine positioning, and medical staff should be aware of the importance of avoiding excessive extension and flexion during anaesthetic, surgical or radiological interventions.[6] This advice should be repeated at surveillance appointments and during examinations for myelopathic signs.[6] Asymptomatic children should be advised to avoid certain activities such as football, gymnastics and trampolining under the age of six years.[6] A symptomatic child should have plain cervical spine X-rays undertaken whilst in the neutral position.[6] If there are no radiographic changes, then flexion–extension views should be sought and a referral to a spinal surgeon made.[6]

A large tongue and a restricted mouth opening can make maintaining the airway difficult. Often, a jaw thrust is performed gently, to avoid subluxation of the temporomandibular joint.[8] Endotracheal tube size may be smaller than anticipated because of the presence of subglottic stenosis. In 2008, the National Institute for Health and Care Excellence published guidance regarding the best clinical practice for prophylaxis against infective endocarditis in patients with pre-existing cardiac lesions undergoing certain interventional procedures.[24] They advised not offering antibiotic prophylaxis in these patients, but explaining the importance of maintaining good oral health.

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Anaesthesia-related complications can include severe bradycardia, airway obstruction, post-intubation stridor and bronchospasm.[8,25]

Down's syndrome is associated with premature ageing. The prevalence of age-related conditions, including visual and hearing defects, epilepsy, thyroid disorders, and dementia, sharply rises after the age of 40 years.[26] Scal[27] and others[28] have reported a reluctance for families and paediatric practitioners to initiate the transition to adult-based services, and in some cases there was a preference to remain with paediatric services. Jensen and Davis found that half of the individuals with Down's syndrome who were investigated continued to use child-focused providers.[29]

As patients with Down's syndrome are living longer, their needs for surgery and anaesthesia increase. It is therefore imperative that surgeons and anaesthetists are well versed with this condition. Awareness should also be raised within the training programmes for doctors and allied professionals. De-institutionalisation in developed countries of patients with Down's syndrome has inadvertently led to reduced access to experienced health professionals and specialists, with more strain on carers and general practitioners.[4] Greater focus is needed on the prevention and treatment of conditions that develop in middle and older age. These patients should be viewed as having the likelihood of a good clinical outcome and sustained quality of life.[4]

- **The ENT manifestations of Down's syndrome are well documented**
- **Our study shows that ENT procedures for head and neck manifestations are dominant in Down's syndrome children**
- **Close links between the otolaryngologist, cardiac surgeon, general surgeon and paediatricians are required to ensure coherent and transparent care**
- **Swallowing dysfunction and delayed phonological development necessitates early, continued input from speech therapists, dysphagia professionals and audiologists**
- **The longer life expectancy of Down's syndrome patients necessitates greater emphasis on prevention and treatment of conditions**
- **In addition, better streamlining of paediatric to adult care is required**

Appropriate care of children with Down's syndrome requires subspecialty and multidisciplinary input. The provision of optimal care at home for a child with Down's syndrome poses a significant dilemma. Such care often starts with referral for specialised evaluation within the context of their home, followed by close communication between tertiary and primary care providers.[2] The Down's Syndrome Association website is an excellent point of information for patients, carers and health professionals.[3] There is an annual health checklist that provides a guide for general practitioners to manage these patients in the community.[3]

In conclusion, the life expectancy of patients with Down's syndrome has overall increased from 25 to 60 years. ENT manifestations of Down's syndrome are common. Hence, greater provisions are needed to streamline otolaryngology services for children and for better transition of care to adult services.

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