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Otorhinolaryngological manifestations in Down syndrome: MERF experience

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ABSTRACT

Background: Syndromic children often pose a spectrum of challenges to the paediatric otolaryngologist. Syndromes like the Down syndrome present with a multitude of manifestations in the head and neck region and require careful management.
Methods: A retrospective study of thirty patients with Down syndrome presenting from January 2013 to January 2018 in a tertiary care setting was done. All patients were evaluated by ENT examination and further investigations based on the complaints. The aim was to study the various ENT disorders in Down syndrome patients and their management.
Results: The most common disorder was hearing loss in eleven patients followed by rhinosinusitis in ten patients, adenoid hypertrophy and otitis media with effusion (OME) in six patients and obstructive sleep apnea in three patients. Most of the patients had medical management; very few needed surgery. Successful outcomes were possible for all patients.
Conclusions: Dealing with otorhinolaryngological disorders may pose a management challenge in Down syndrome and this requires a well experienced ENT, medical and rehabilitative team to provide optimal management and good quality of life to these children.

Keywords: Down syndrome, Adenoid hypertrophy, Obstructive sleep apnea, Hearing loss

INTRODUCTION

Down syndrome (DS) is one of the most common chromosomal disorders in children and has an incidence of 1 in 700. The quality of life and development is affected in children with DS as they are predisposed to a number of medical problems.[1] Several ENT and systemic manifestations have been described and appropriate and early intervention is important. Hearing loss is prevalent in children with DS and is found to be 38–78%.[2] Patients with Down syndrome have increased risk of obstructive sleep apnea. Awareness of the ENT disorders in DS is vital for optimal outcomes. The aim of this study was to study the various otorhinolaryngological disorders in Down syndrome patients and their management in a tertiary care centre.

METHODS

A retrospective study of thirty patients with Down syndrome presenting from January 2013 to January 2018 was done in Madras ENT Research Foundation, Chennai, India. Ethical approval for the study was taken from the Institutional review committee. Inclusion criteria included patients with ENT disorders in Down syndrome. Patients who had syndromes other than Down syndrome

were excluded from the study. All patients were evaluated by ENT examination and further investigations based on the complaints. The various ENT disorders were investigated and found to be hearing loss (conductive/sensori-neural/mixed hearing loss), otitis media with effusion, adenoid hypertrophy, rhinosinusitis and obstructive sleep apnoea. All these patients were treated by appropriate medical management and a few of these patients required surgery. For the statistical analysis, Student's t test was used. The follow-up period in our cohort ranged from 8 months to 3 years and the mean follow-up period was 24 months.

RESULTS

A retrospective study of thirty patients with Down syndrome was done over a five-year period in a tertiary care hospital. There were eighteen males and twelve females in the study and the age range of the patients was 3 to 35 years (mean, 11 years) (Figure 1). The most common disorder (Figure 2) was hearing loss in eleven patients (37%) followed by rhinosinusitis in ten patients (33%), adenoid hypertrophy and otitis media with effusion (OME) in six patients (20%) (Figures 3 A,B and 4) and obstructive sleep apnea in three patients (10%). Of the eleven patients with hearing loss, six patients had conductive hearing impairment, two had sensori-neural hearing loss and three patients had mixed hearing loss.

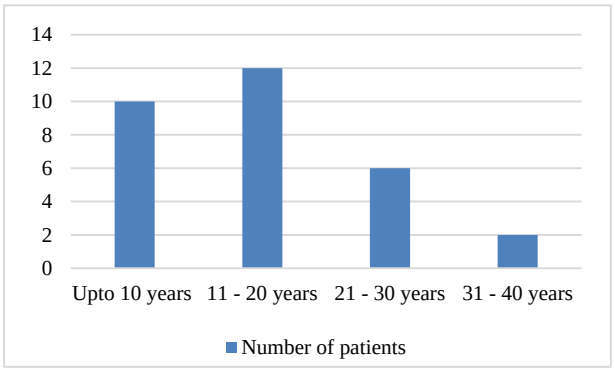

Figure 1: Age range of patients with Down syndrome.

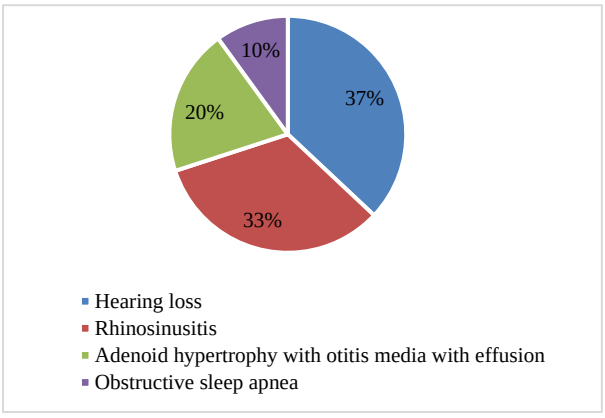

Figure 2: ENT disorders encountered in Down syndrome.

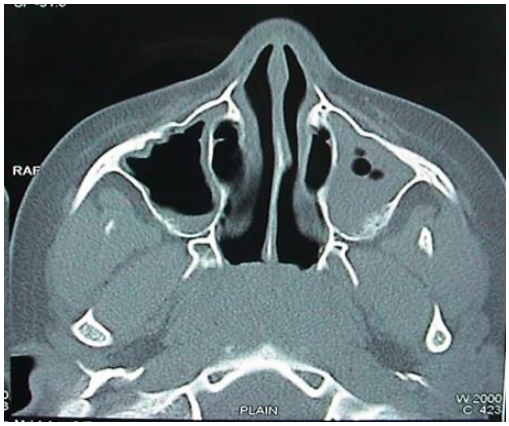

Figure 3A: Adenoid hypertrophy and rhinosinusitis in a child with Down syndrome.

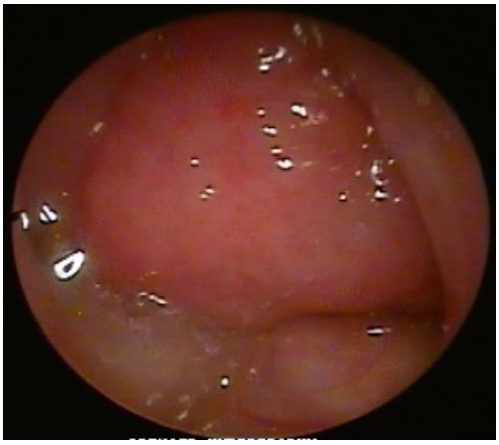

Figure 3B: Adenoid hypertrophy in a child with Down syndrome.

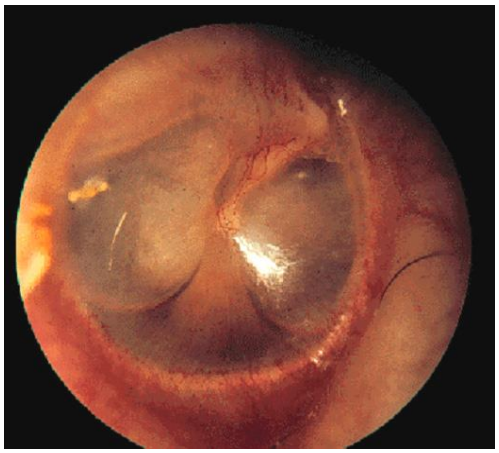

Figure 4: Otitis media with effusion in Down syndrome.

Patients with hearing loss were managed depending on the aetiology. Six patients with conductive hearing loss due to OME were treated by medical management; three of them required placement of grommets. In all six patients with conductive hearing loss, the post-operative pure tone audiometry after one month showed normal

hearing which was statistically significant ($p<0.05$) and confirmed that medical/surgical treatment of otitis media with effusion is effective in correcting conductive hearing loss. In our study, two patients with sensori-neural hearing loss were advised hearing aids. Three patients with mixed hearing loss were treated medically for OME and did not require hearing aids. Patients with rhinosinusitis, adenoid hypertrophy and otitis media with effusion were managed medically. Thirty percent of patients underwent surgical management. Six children with enlarged adenoids and otitis media with effusion underwent adenoidectomy and bilateral myringotomy and grommet placement (20%). There were three patients with obstructive sleep apnea due to adenotonsillar hypertrophy and symptoms resolved with adenotonsillectomy (10%) (Figure 5).

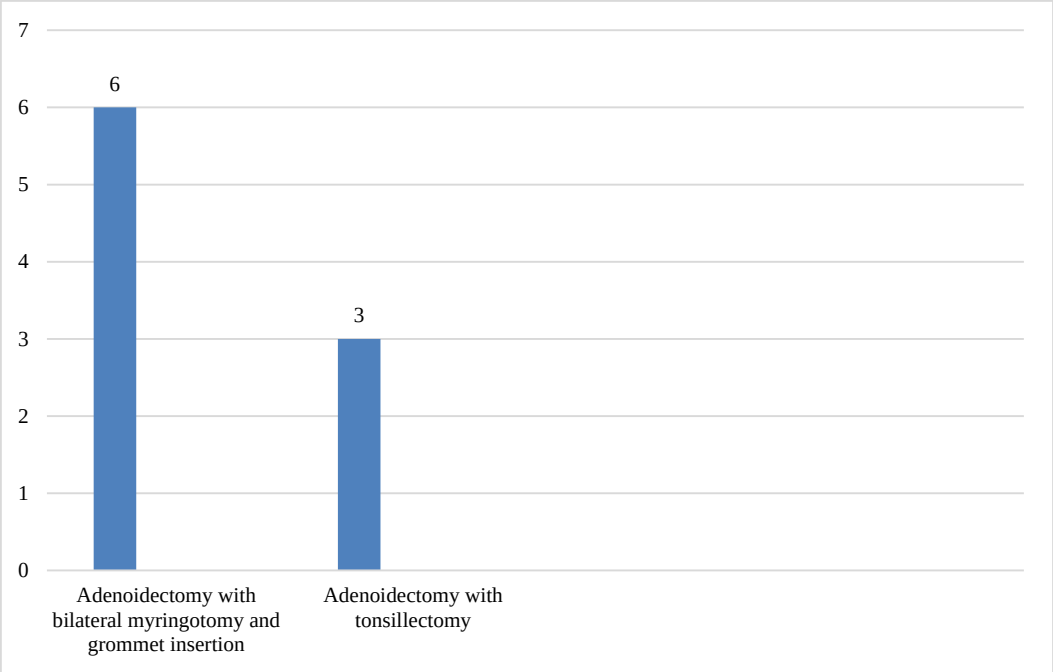

Figure 5: Surgical management of Down syndrome.

All patients had successful management of their disease. In the patients managed surgically, there were no intra-operative or post-op complications in any of the patients. Overall there was no recurrence of ENT disease in our cohort of medically and surgically managed patients over their follow up period which ranged from 8 months to 3 years with the mean follow-up period being 24 months.

DISCUSSION

Down syndrome patients are prone to several otorhinolaryngological disorders. Knowledge of the presenting features, prevalence and management protocol of ENT conditions in DS children is important. In this study, eleven patients (37%) were diagnosed with hearing loss of which six patients had conductive hearing impairment, two had sensori-neural hearing loss and three patients had mixed hearing loss. In the literature, the prevalence of hearing loss in children with DS is found to be 38–78%.[2] Conductive hearing impairment is common in view of the common association with various structural anomalies. These include microtia, external auditory canal stenosis predisposing to cerumen impaction, midface hypoplasia and constricted eustachian tubes openings. Eustachian tube collapse due to generalized hypotonia causes dysfunction of tensor veli palatini muscle of palate which leads to increased prevalence of acute otitis media and chronic otitis media with effusion. Others factors which contribute to conductive hearing impairment include recurrent upper respiratory tract infections due to impaired immune function and malformed ossicular chain.[3,4] Increased risk of cholesteatoma has been reported. There is anecdotal evidence of increased congenital sensorineural hearing loss (SNHL) amongst this population. The incidence of SNHL discovered on newborn hearing screening in Down syndrome is 4%.[5,6] The various causes of SNHL are reduced spiral ganglion cells, shortened organ of Corti, Mondini dysplasia and compression of the auditory nerve in the internal auditory meatus leading to nerve degeneration. Sensorineural deficit occurs earlier when compared with normal population in later life of DS children suggesting early presbycusis.[3,7]

Shott et al demonstrated that with early medical and surgical interventions of chronic otitis media in young DS children, 98% attained normal hearing after treatment which is crucial to language development.[8] DS children have poor attention and inconsistent response which makes behavioural testing and hearing evaluation difficult. Brainstem evoked response audiometry is a

useful objective test, which can evaluate auditory performance regardless of the patient's age and mental status, particularly in young and uncooperative children who are not suitable for behavioural audiogram or pure tone audiometry.[9]

Otitis media with effusion is managed medically or surgically. In our study, six children with otitis media with effusion required placement of grommets. Often multiple sittings of grommet insertion will be required.[10] Hearing loss in DS is managed by hearing aids but hearing aid fitting is challenging due to behavioural problems and narrow ear canals.[10] Narrow external auditory canals may result in wax impaction which can compromise hearing and cause feedback from hearing aids. If recurrent ear infections prevent the use of conventional hearing aids, bone conduction or bone anchored hearing aids (BAHA) should be considered.[11]

Respiratory tract infection is the most common cause of hospitalization in Down syndrome children less than 3 years of age with 80% occurring before 2 years of age.[12,13] Multi-factorial aetiology such as adenoid hypertrophy, sinus infection and allergy due to low immunity predispose to chronic rhinosinusitis in DS children.[14] The hypoplasia of the skeletal structure of the mid-face tends to limit the space of the nasal passages, nasopharynx and oropharynx. This leads to mucus pooling and stagnation within the nasal passages, which provides for a focus for infection. Treatment includes nasal saline douches, encouraging nose blowing, oral anti-histamines, nasal or oral decongestants and antibiotics. Adenoidectomy has a role in some children. Children with DS also have a higher incidence of laryngopharyngeal reflux, which can contribute to nasal and throat symptoms. Symptoms of gastro-oesophageal reflux can be managed with H2-receptor antagonists.[15]

Obstructive sleep apnea was seen in three of our patients (10%). OSA can affect up to 50% of the paediatric DS population, with >90% of DS adults manifesting some aspect of OSA.[16] The prevalence of OSA in children with DS ranges between 30% and 60% when compared to the general paediatric population with a range between 0.7% to 2%. Multiple and complex factors predispose children with DS to develop airway obstruction and subsequently OSA. Anatomical abnormalities such as narrow nasopharynx, midfacial and mandibular hypoplasia, adenotonsillar hypertrophy, macroglossia (due to crowding of the oropharynx) and a shortened palate along with generalized hypotonia, thyroid dysfunction, immature immune system (predispose to recurrent respiratory infections), and a propensity for obesity predispose DS children to upper airway obstruction. Associated conditions such as chronic lung disease and gastroesophageal reflux may worsen airway problems.[17] The 2011 "Health Supervision for Children with Down Syndrome" clinical report by Bull and the American Academy of Paediatrics Committee on Genetics recommends discussing the symptoms of sleep difficulties with parents and that all children must undergo an overnight diagnostic polysomnography by 4 years of age.[18]

Pharmacological treatments for OSA have limited safety and efficacy data for children with DS. Adenotonsillectomy can produce significant benefit for younger children with OSA, but nocturnal hypoxia can persist despite surgery in many cases or it can return in later childhood.[2] Lingual tonsil hypertrophy is more common in refractory OSA children with DS who have undergone adenotonsillectomy previously.[19] Lingual tonsil ablation can produce significant improvement in the airway in selected cases. Management of refractory OSA in children where surgical remedies have been exhausted includes positive pressure ventilation in the form of continuous positive airway pressure or bi-level positive airway.[20]

Subglottic and tracheal stenosis, laryngomalacia and vocal cord paralysis are not infrequently seen in the DS patient.[19] Modification of the standard intubation technique may be required when subglottic stenosis is present in a child with DS. Endotracheal tubes that are at least two sizes smaller than what is appropriate for the patient's age must be used. Prognosis of subglottic stenosis in DS children is poor as lower success rates of decannulation are reported and also repair of stenosis becomes more difficult.[21]

Recurrent multiple cervical lymphadenitis has been reported in DS. Increased prevalence of both congenital hypothyroidism (CH) and acquired thyroid dysfunction have been reported in DS children. The most common thyroid dysfunction profile seen in DS children is mild elevation of plasma thyrotropin (TSH) with normal thyroxine (T4) levels.[22] The most frequent genetic causes of intellectual disability and the most frequent genetic cause of mental retardation in children is DS.[23] Autoimmune diseases such as Hashimoto's thyroiditis (HT), graves' disease (GD), alopecia, vitiligo, type 1 diabetes, celiac disease and idiopathic arthritis are more common. There is an increased risk of type 1 diabetes mellitus, and haematological malignancies.[24]

A variety of ENT surgeries may need to be performed in DS children and the co-morbidities associated with DS are to be considered prior to taking up these children under general anaesthesia. Congenital heart disease (CHD) has a prevalence of 40 to 63.5% in DS.[25] Atlantooccipital instability affects up to 30% of patients with Down syndrome. Specific care must be taken with these patients as hyperextension or hyper-rotation of the neck, especially during ENT surgery (adenotonsillectomy and ear procedures), could cause damage to the spinal cord.[26] Anaesthetists should be aware of the increased risk of intubation bradycardia, loss of the airway on induction because of adenotonsillar hypertrophy and poor tone, and difficult intubation because of a large tongue. Post-operatively, these patients may have difficulty in

maintaining their airway because of hypotonia and close observation in recovery is recommended. Post-extubation stridor is significantly higher in children with DS.[27] Iron-deficiency anaemia (10%), leukaemia (1%), coeliac disease (5%), immune dysfunction, diabetes mellitus (1%) and Hirschprung's disease are more common in children with DS which may also influence their anaesthesia.[25] In our study, none of our patients had an anesthetic complication because of a thorough pre-operative assessment, intra-operative and post-operative monitoring by an experienced anesthesia team.

In our study, the common ENT conditions encountered in Down syndrome included hearing impairment, rhinosinusitis, adenoid hypertrophy, otitis media with effusion and obstructive sleep apnea. Following a management protocol for all patients with Down syndrome helped ensure good outcomes.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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