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Adenotonsillectomy should be avoided whenever possible in infantile-onset Pompe disease

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Pompe disease is a lysosomal storage disorder caused by acid alpha-glucosidase deficiency. Classic infantile-onset Pompe disease (IPD) is characterized by cardiomyopathy, hypotonia, respiratory insufficiency, and death from cardiorespiratory failure by age 2 years. Enzyme replacement therapy (ERT) with alglucosidase alfa (Genzyme, Cambridge, MA) has improved clinical outcomes. However, as we establish the natural history of long-term survivors of IPD, new clinical challenges emerge.

Hypernasal speech and velopharyngeal incompetence is a prominent feature of IPD and among the primary clinical challenges faced by this population [1–4]. Hypernasality is a perceptual phenomenon in which speech sounds disproportionately nasal due to excess sound resonating in the nasal cavities during the production of oral sounds [5]. Hypernasality is often severe and persistent in children with IPD, even when treated early with long-term ERT and despite speech therapy [1,3,4]. Velopharyngeal incompetence in IPD may be compounded by involvement of other bulbar muscles which may contribute to dysphonia, disordered articulation, feeding difficulties, and dysphagia [1,3,4,6].

Adenotonsillectomy (AT) is performed commonly in children for recurrent throat infection and sleep disordered breathing (SDB) [7]. Hypernasality is a well-known risk associated with adenoidectomy [8–12]. Velopharyngeal closure is achieved in most children via velo-adenoidal closure; adenoidectomy effectively deepens the nasopharynx increasing the distance needed for velar closure [13]. Permanent hypernasality post-adenoidectomy occurs in approximately 1:1500 cases [12,14]. Risk factors include submucous cleft palate, preoperative hypernasality, and neuromuscular disorders [8–12].

AT is considered in patients with IPD due to recurrent throat infection and SDB. As these patients often have preoperative hypernasality due to the underlying disease they are at increased risk for worsening of hypernasality post-AT. Therefore, we recommend that AT in patients with IPD be avoided when possible. Alternative approaches to manage SDB including non-invasive positive pressure ventilation, positional therapy, tonsillectomy alone, and/or trial of nasal corticosteroids should be considered before adenotonsillectomy in this high-risk population.

Declaration of Competing Interest

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