22q11.2 Deletion syndrome: What the ENT surgeon needs to know about Velopharyngeal Insufficiency and its impact on Speech and Language Development.

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Background

22q11.2 Deletion syndrome (22q11.2DS) occurs in 1 in 4000 live births [1] and often results in significant speech and language delay [1,2]. This is due to delay in both language and speech acquisition; the latter often contributed to by the presence of submucosal cleft palate (SMCP) and velopharyngeal insufficiency (VPI) [1]. Other features of 22q11.2DS include learning difficulties, muscle hypotonia and hypoplasia and laryngeal abnormalities, all of which can contribute to poor speech acquisition [2].

Objectives

To compare speech outcomes between different surgical techniques for SMCP and VPI repair in 22q11.2DS and non-syndromic children following cleft palate (CP) repair.

Methods

Literature search of surgical techniques for SMCP repair and VPI surgery in 22q11.2DS. Comparison of speech outcomes for SMCP and VPI in 22q11.2DS and CP repair in non-syndromic CP.

Results

In 22q11.2DS, palatoplasty alone results in normal voice resonance in only 20% of patients [1]. Soft palate movements tend to be described as sluggish and poorly co-ordinated. Half of the remaining patients require further pharyngoplasty surgery [1,3]. Primary pharyngoplasty surgery results in normal resonance in 40-60% with further surgery required in 10-13% [1,3]. In non-syndromic CP, palate repair before 12 months results in normal voice at 5 years in 48+/-3% and speech with no evidence of structural palatal problems in 66+/-3%. Secondary surgery is reported in 20% and ongoing palatal speech issues in 17% [4].

Figure 1 (left).
Rest image of a 4-year old child with 22q11.2DS demonstrating a relatively unremarkable palate with a typical large nasopharynx.

Figure 2 (left).
Shot of maximum attempted closure in the same child as Figure 1. There is slight thinning of the anterior soft palate, a poorly defined knee (which is anterior to the ideal lift point) and a large velopharyngeal gap.

Conclusions

Speech outcomes are worse and secondary surgery more common in children with 22q11.2DS compared with non-syndromic CP [1,3,4]. Hypotonia/muscle hypoplasia and subtle laryngeal abnormalities potentially contribute to poor post-operative speech outcomes in 22q11.2DS [2].

References

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Authors declare no conflicts of interest in the production of this poster.

Special thanks to Mr Jonathan Syme-Grant, Queen's Medical Centre, Nottingham, UK, for the acquisition of images used in this poster.