7q11.23 duplication syndrome: Challenges of ear surgery for cholesteatoma management

Dioni Petsiou[1], Marcel Geyer[2]

[1]School of Medicine, National and Kapodistrian University of Athens, Athens, Greece
[2]ENT Department, Salisbury NHS Foundation Trust, Salisbury, United Kingdom

INTRODUCTION

7q11.23 duplication syndrome (DS) is a rare chromosomal anomaly, resulting from an additional copy of a portion of the long arm of chromosome 7. The estimated prevalence is one in 7,500 – 20,000 individuals. The inheritance transmission pattern is autosomal dominant. Limited literature information on the otolaryngological manifestations of the syndrome includes include chronic otitis media, hearing loss and obstructive sleep apnea.

CASE DESCRIPTION

A 9-year-old female diagnosed with 7q11.23 DS was referred to our ENT outpatient clinic with a persistently discharging right ear and a history of chronic recurrent right ear infections.

Clinical examination
Copious thick right ear discharge and inflammation that made identification of the tympanic membrane (TM) difficult.

Diagnostic investigations
Tympanometry: Type B (flat) impedance traces with normal canal volume
Audiogram: Moderate conductive hearing loss in both ears (right ear thresholds between 30-60 dB).
Temporal bone CT scan:

- Opacification of the right external auditory meatus, the middle ear cavity and the mastoid.
- Proximity of the jugular foramen to the external auditory meatus and tympanic annulus posteriorly.
- Bilaterally the posterior attachment of the TM (annulus) abutted the bony contour of the jugular foramen and dehiscent jugular bulb.

Management

- Suggested diagnosis: right ear cholesteatoma
- Combined – approach tympanoplasty operation
- During the surgery she was found to have a dehiscent jugular bulb adherent to the tympanic membrane and low dura making access difficult.
- Surgery carried without complications, removing the cholesteatoma from the middle ear.
- Post – operative audiogram (3 months later): deterioration in the right ear thresholds to 50-75 dB.

FIGURES

AUDIOGRAMS

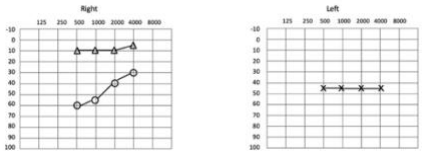

Figure 1. Pre-op audiogram

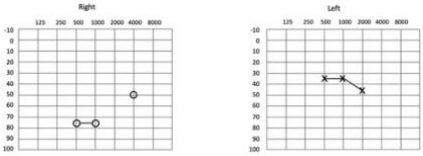

Figure 2. Post-op audiogram

CT SCANS

A) Axial cuts

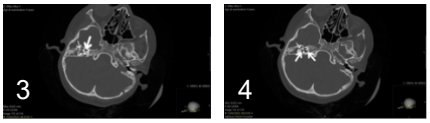

Figure 3. Normal right middle and inner ear structures (ossicles and cochlea). *Thick arrow* cochlea. *Thin arrow* head of the malleus.

Figure 4. *Thick arrows* the bony contours of the right inner ear structures are intact. *Thin arrow* horizontal portion of the facial nerve.

B) Coronal cuts

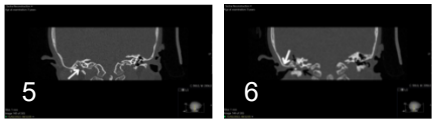

Figure 5. *White arrow* opacification of the middle ear cavity and possible incus erosion

Figure 6. *White thick arrow* low tegment and dura. *White thin arrow* lateral semicircular canal. *Black thick arrow* high riding jugular bulb. *Black thin arrow* opacified epitympanum. *Asterisk* opacified external rigt ear canal.

DISCUSSION

- There is a lack of information available discussing clinical details and results for 7q11.23 DS patients, including details of otoscopy, tympanometry, air and bone conduction hearing tests, behavioural testing and distortion product otoacoustic emissions (DPOAEs)
- Data is available for Williams – Beuren Syndrome (WS), a genetic disorder caused by a heterozygous microdeletion of chromosome 7q11.23
- Individuals with WS and "normal" hearing tend to have outer hair – cell dysfunction around the frequency of 4000 Hz in comparison to normal – hearing controls.
- WS patients commonly present with chronic middle ear infections and TM retraction, which was also seen in our case.
- The surgical risks of the combined – approach tympanoplasty procedure is overall accepted as low. However, our patient was susceptible for additional potentially significant complications, due to the fact that she has a right – sided high and dehiscent jugular bulb as well as low tegmen. In patients with dehiscent jugular bulb, significant bleeding might occur during myringotomy, tympanomeatal flap elevation and middle ear surgery. Persistent haemorrhage is usually controlled by gauze – packing or Gelfoam application in the external auditory meatus. A rarely used alternative option would be to use transcatheter endovascular embolization. The last resort in controlling the haemorrhage should be to ligate the internal jugular vein, eradicating the sigmoid sinus.

CONCLUSION

The otolaryngological manifestations of 7q11.23 DS are complex and poorly described by the literature. Hopefully, future clinical studies will expand our knowledge and contribute to the better understanding of the profile and management options for such rare cases.

REFERENCES

(1) Osborne LR, Mervis CB. 7q11.23 deletion and duplication. Vol. 68, Current Opinion in Genetics and Development. Elsevier Ltd; 2021. p. 41–8.
(2) Mervis CB, Morris CA, Klein-Tasman BP. 23 Duplication Syndrome. 2015.
(3) Dixit A, McKee S, Mansour S, Mehta SG, Tanteles GA, Anastasiadou V, et al. 7q11.23 Microduplication: A recognizable phenotype. Clin Genet. 2013 Feb;83(2):155–61.
(4) Morris CA, Mervis CB, Paciorkowski AP, Abdul-Rahman O, Dugan SL, Rope AF, et al. 7q11.23 Duplication syndrome: Physical characteristics and natural history. Am J Med Genet A. 2015 Dec 1;167(12):2916–35.
(5) Marler JA, Sitcovsky JL, Mervis CB, Kistler DJ, Wightman FL. Auditory function and hearing loss in children and adults with Williams syndrome: Cochlear impairment in individuals with otherwise normal hearing. Am J Med Genet C Semin Med Genet. 2010 May 15;154(2):249–65.
(6) Oliveira MW, Lavinsky J, Valerio MMH, Felix TM, Lavinsky L. Tympanic Membrane Retractions in patients with Williams Syndrome: A Controlled Study. Int Arch Otorhinolaryngol. 2020;
(7) Goodey RJ, Smyth GDL. Combined Approach Tympanoplasty in Children. Laryngoscope. 1972;82(2).
(8) Atmaca S, Elmali M, Kucuk H. High and dehiscent jugular bulb: Clear and present danger during middle ear surgery. Surgical and Radiologic Anatomy. 2014;36(4):369–74.
(9) Fox R, Nash R, Tatla T. Encountering a high jugular bulb during ear surgery. Ann R Coll Surg Engl. 2017;99(1):e36–7.