Congenital Deafness with Ear Deformities in Arthrogryposis Multiplex Congenita (AMC)

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Introduction

First described by Otto in 1841, arthrogryposis multiplex congenital (AMC) is a rare disorder currently listed under the National Organisation of Rare Disorders (1). It is characterized by joint contractures that are present at birth in at least two areas of the body. The authors report an uncommon case of associated congenital deafness with ear deformity in a patient with AMC.

Methods

54 year old female patient with bilateral congenital profound mixed hearing loss requiring hearing aids from birth was reviewed with the otoendoscope revealing a number of otological deformities including low set ears, deformed pinna, deformed external auditory canal and possible middle ear deformity. CT-temporal bones revealed upslanting external auditory canals but no overt deformity of the ossicles. The patient was managed conservatively after exclusion of evidence of cholesteatoma on radiological imaging.

Right ear

Left ear

Results

Only one other case with associated middle ear deformity in AMC has been reported in the literature (2).

Conclusion

Associated congenital deafness and ear deformities are possible features in patients with AMC that the otorhinolaryngologist should include in their clinical repertoire.

References

1. Otto AW, Peltier LF. A Human Monster with Inwardly Curved Extremities. Clinical Orthopaedics and Related Research. 1985 Apr 1;194:4-5.
2. Quinn SJ, Bleach NR, Richards AE. Middle ear deformity in arthrogryposis multiplex congenita. Annals of Otology, Rhinology & Laryngology. 1994 Sep; 103(9):729-31.