

Case Report

Axial Mesodermal Dysplasia Complex with a Unique Abnormal Course of Vestibulocochlear Nerve

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Axial mesodermal dysplasia complex (AMDC) is a combination of multiple congenital malformations arising due to the mesodermal cell migration, neural tube fusion, and rhombencephalon segmentation. Here, we present the imaging findings of a 15-year-old boy with AMDC who has bilateral inner ear malformations associated with a vestibulocochlear nerve extending to Meckel cave, cystic lesion in prepontine cisterna, cervical vertebral segmentation anomalies, and maxillary bone anomalies.

KEYWORDS: Axial mesodermal dysplasia complex, computed tomography, magnetic resonance

INTRODUCTION

Axial mesodermal dysplasia complex (AMDC) is a combination of various congenital malformations including the cardiovascular, pulmonary, gastrointestinal, genitourinary, musculoskeletal, and central nervous system abnormalities. AMDC also includes many syndromes such as vertebral, anorectal, cardiac anomalies, tracheo-esophageal fistula, esophageal atresia, renal and limb anomalies (VACTERL), Müllerian duct aplasia, renal agenesis, cervicothoracic somite dysplasia (MURCS), Goldenhar (oculo-auriculo-vertebral spectrum), and Klippel-Feil (cervical vertebra fusion syndrome) ^[1].

Here in, imaging findings of a 15-year-old boy with AMDC who has bilateral inner ear malformations associated with vestibulocochlear nerve extending to Meckel cave, cervical vertebral segmentation anomalies, maxillary bone anomalies, and cystic lesion in prepontinecistern are presented.

CASE PRESENTATION

A 15-year-old boy with bilateral congenital hearing loss and congenital cervical scoliosis was admitted to our ENT Department. Physical and audio-logic examination revealed cervical scoliosis, facial asymmetry, bilateral sensorineural hearing loss, and right nervusabducens paralysis. Temporal bone computed tomography (CT; Toshiba Aquilion 64, Toshiba Medical Systems, Tokyo, Japan) demonstrated the absence of the normal differentiation of the cochlea and vestibule which was replaced by a cystic structure in the left side. These features were classic examples of common cavity malformation (Figure 1). Additionally, CT revealed a right cystic cochlea lacking the entire modiolus, along with an enlarged cystic vestibule. Internal auditory canal (IAC) was hypoplastic on both sides. These features were components of incomplete partition type I (Figure 2) ^[2]. Inner ear magnetic resonance imaging (MRI) T2-SPACE (Signa Excite, GE Medical Systems, Milwaukee, WI, USA) sequence showed the left vestibulocochlear nerve coursing toward Meckel cave (Figure 3 a-c). The left cochlear, superior and inferior vestibular nerves were seen entering the Meckel cave. The left trigeminal nerve and Meckel cave were smaller than the right trigeminal nerve (Figure 4). The left facial nerve, the right vestibulocochlear nerve, and the right facial nerve were seen at normal location. The right abducens nerve was not demonstrated due to aplasia. Left abducens nerve was found normal. A cystic lesion was seen in the prepontine cisterna (Figure 5).

Cervical radiography showed cervical vertebrae segmentation anomalies at multiple levels, cervical scoliosis, and maxillary bone anomalies (Figure 6).

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Abdominal ultrasound, which was performed to evaluate kidney anomaly, was normal.

Based on these clinical and imaging findings, the patient was finally diagnosed with AMDC. This case report is presented with the consent from the patient's parent.

DISCUSSION

Axial mesodermal dysplasia complex is an extremely variable condition. Deformities associated with AMDC are raised due to the mesodermal cell migration, neural tube fusion, and rhombencephalon segmentation. Most of the AMDC cases are of sporadic nature. However, the causes are unclear [1-3].

Our patient showed multiple inner ear anomalies, left trigeminal nerve hypoplasia, aberrant course of left vestibulocochlear nerve,

right nervusabducens aplasia, cervical vertebral segmentation anomalies, maxillary bone anomalies, and cystic lesion in prepontine cisterna.

Anomalies of the vestibulocochlear and facial nerves in the spectrum of hypoplasia and aplasia are well described with ear anomalies. Aberrant course of the facial nerve is also well described with ear anomalies [4-5]. However, we could not find a case with aberrant course of the vestibulocochlear nerve entering the Meckel cave on PubMed (MEDLINE) and Google scholar in English literature. Only exception was the electronic presentation (online) by Wang et al. who described three cases with bilateral hearing loss without any syndrome [6]. In two of these cases, vestibulocochlear nerves were seen coursing toward Meckel cave on MRI. In the third case, the superior vestibulocochlear nerve was identified more superiorly,

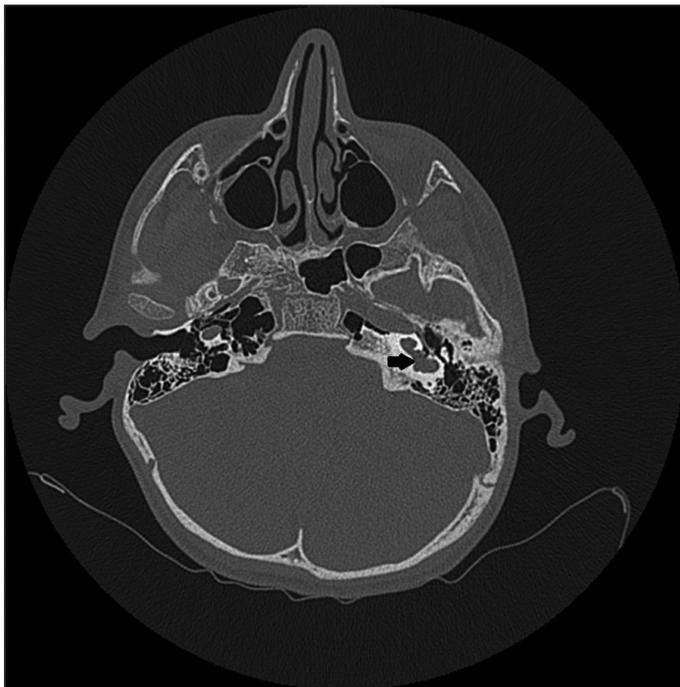


Figure 1. Temporal bone CT displays common cavity malformation in the left ear.

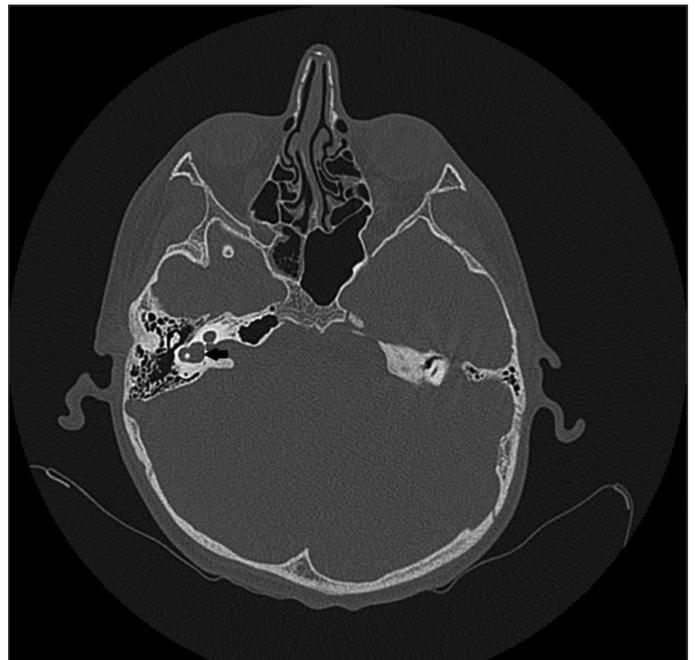


Figure 2. Temporal bone CT shows findings consistent with incomplete partition type 1 including a right cystic cochlea lacking the entire modiolus, along with an enlarged cystic vestibule. IAC was deficient in the right side.

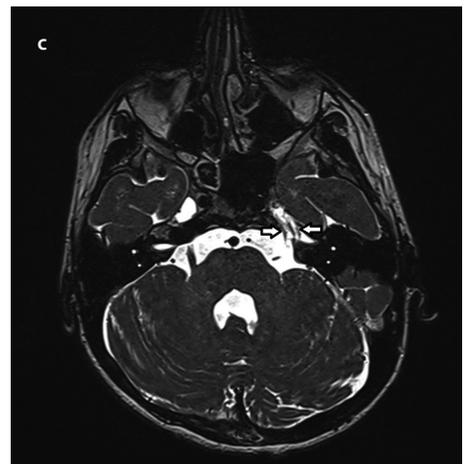
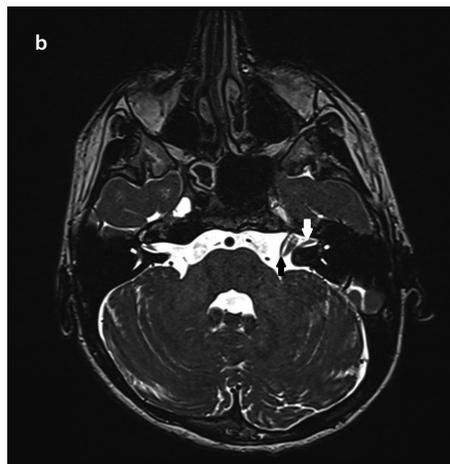
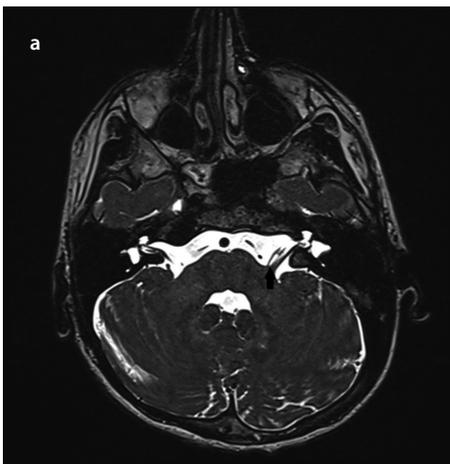


Figure 3. a-c. Inner ear MRI with T2-SPACE sequence shows facial nerve and vestibulocochlear nerve (black arrow) emerging from the normal localization on brain stem (a). The left vestibulocochlear nerve (black arrow) coursing toward Meckel cave and facial nerve extending to the IAC (white arrow) (b). The left cochlear, superior and inferior vestibular nerves were seen at the entry of Meckel cave (white arrows) (c).

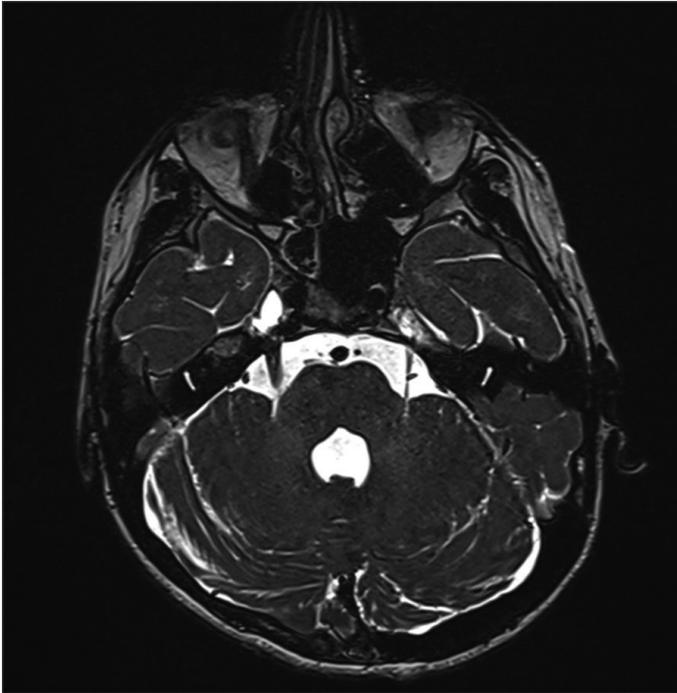


Figure 4. MRI shows the smaller trigeminal nerve and Meckel cave in the left side when compared to the opposite side.

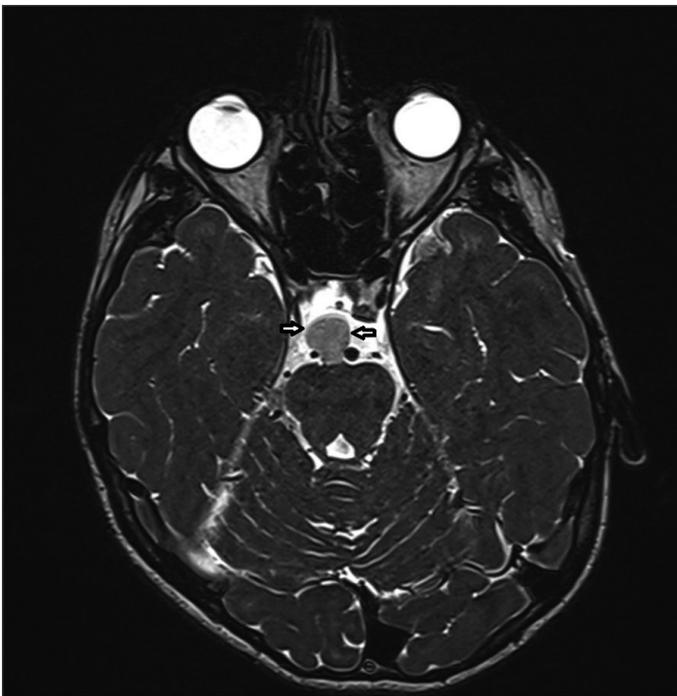


Figure 5. A cystic lesion (arrows) is seen in prepontine cisterna.

coursing toward the lowermost aspect of Meckel cave, inferior to the right trigeminal nerve. The facial nerve was identified in the anterosuperior part of the IAC. A small nerve was also identified in the posterior IAC, which likely represents the inferior vestibular nerve. No cochlear nerve was identified. IACs were hypoplastic/atretic in two of these cases, and one mandibular was smaller [6]. Our case showed the left vestibulocochlear nerve coursing toward Meckel cave. The left trigeminal nerve and Meckel cave were smaller than the right. Right abducens nerve was aplasia. Left common cavity



Figure 6. Cervical radiography shows multilevel cervical vertebral segmentation anomalies and maxillary bone anomalies.

malformation and right incomplete partition type I was demonstrated. Additionally, we demonstrated prepontine cystic lesion, right abducens nerve aplasia, cervical vertebral segmentation anomalies, and maxillary bone anomalies.

Normal development of the cranial nerve nuclei is derived from 2-3 and 4-5. rhombomeres. The neuronal precursors display programmed migratory behavior and send axons along defined trajectories to their peripheral targets. Metencephalon contains neurons of the trochlear, trigeminal, abducens, facial, and vestibulocochlear nerves. The facial cartilages and bones are derived from branchial arches, mesodermal structures, and paraxial mesoderm. Facial structures and cranial sensory nerves are arising from the neural crest cells [7]. AMDC arises due to the mesodermal cell migration, neural tube fusion, and rhombencephalon segmentation. So, the cardiovascular, pulmonary, gastrointestinal, genitourinary, musculoskeletal, and central nervous system anomalies are defined in AMDC [1-3]. Our patient showed multiple inner ear and skeletal anomalies.

Goldenhar syndrome is a rare congenital malformation involving-
pibulbarderoids, auricular deformities, lid colobomas, hemifacial-
microsomia, facial asymmetry, and vertebral anomalies^[8]. There was
no ocular anomaly in our patient, a feature that makes diagnosis of
Goldenhar syndrome unlikely.

We presented a 15-year-old boy who showed bilateral sensorineural
hearing loss associated with an aberrant course of vestibulocochlear
nerve, additional cranial nerve anomalies, inner ear anomalies, and ver-
tebral and facial anomalies. These features were consistent with AMDC.

CONCLUSION

The presence of cranial nerve anomalies is wellknown in patients
with inner ear malformations, especially in the setting of a syndrome
such as Goldenhar or axial mesodermal dysplasia complex. However,
associated with the additional findings involving inner ear, cranial
nerves, and skeletal system, there is no report of vestibulocochlear
nerve extending toward Meckel cave. This unique manifestation of
vestibulocochlear nerve may improve our understanding regarding
the embryogenesis of cranial nerves and inner ear structures.

Informed Consent: Written inform consent was obtained from the parents' of
the patient who participated in this study.

Peer-review: Externally peer-reviewed.

Author contributions: Concept – S.T.U., A.O., K.G.; Design – S.T.U., A.O., M.K.;
Supervision K.G., M.K., Y.B.; Resource – K.G., M.K., Y.B.; Materials – S.T.U., A.O.,
Y.B.; Data Collection and/or Processing –S.T.U., K.G., M.K.; Analysis and/or In-
terpretation – S.T.U., A.O.; Literature Search - K.G., M.K., Y.B.; Writing –S.T.U.,
A.O., K.G.; Critical Reviews – K.G., M.K., Y.B.

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