

Case Report

Hearing assessment in a rare case of Hajdu Cheney syndrome

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ABSTRACT

Hajdu Cheney syndrome is extremely rare autosomal dominant congenital disorder of connective tissue. It may occur due to spontaneous de novo mutation and mutation in NOTCH-2 gene identified recently. Most characteristic features include aero-osteolysis involving phalanges of both hands and feet, osteoporosis, deformities of skull, mandible, spine and other bones, kyphoscoliosis and bone fractures. Rarely in some affected individuals, it causes joint hypermobility, dental problems, hearing loss, heart defects, kidney abnormality like polycystic kidneys, excess body hair and recurrent infections in childhood. It affects many parts of body particularly bones. Treatment is symptomatic. In this case report, we present a case of 14 years male child with features of Hajdu Cheney syndrome with genetic predisposition. Patient presented to ENT clinic with complaint of hearing loss.

Keywords: Hajdu Cheney syndrome, Hearing assessment, Hearing loss

INTRODUCTION

Hajdu-Cheney syndrome is rare autosomal dominant disorder causing acroosteolysis of hands and feet, osteoporosis with fracture bony defects, dental abnormalities, craniofacial changes and short stature.¹⁻⁴ It is a rare disease, less than 100 cases have been reported, but its exact prevalence is not known. Signs and symptoms and severity vary greatly among members of same family. Some clinical features are not present at birth, but become apparent in childhood or later⁵. There is presence of mutations in exon 34, terminal exon of NOTCH-2 in Hajdu Cheney syndrome individuals.⁶⁻⁹ NOTCH-2 is located in chromosome 1, 1p13-p11. Craniofacial abnormality, brachycephaly with prominent occiput, hyperthyroidism, low set ears, cleft palate, high arch palate and short neck are classical features in adult patients. Generalized joint hypermobility, spinal abnormalities including deformities, hyposcoliosis, fractures, basilar invagination are present. Hearing loss, dental abnormalities, premature loss of teeth, long bone

deformities are reported. Cardiovascular defects including ventricular septal defects, patient ducted arteriosis, valvular stenosis and insufficiency were also reported.^{10,11} Respiratory infections are common in childhood. Neurological problems including hydrocephalus, sudden death and central respiratory arrest occur due to basilar invagination. Some present with renal cysts or polycystic kidneys and serpentine fibular polycystic kidney syndrome.^{12,13}

CASE REPORT

A 14 yrs old male child presented to our Ramlal ENT, OPD with complaints of decreased hearing since 5 years. Hearing loss was sudden in onset and was non-progressive. There was no history of ear discharge, earache, tinnitus, itching, vertigo, aural fullness. On local examination, ears were at normal position and normal in shape. External auditory canal and tympanic membrane were within normal limits on both sides. On general physical examination, deformity of distal phalanges of

hands and feet. On radiological evaluation, acroosteolysis of distal phalanges of hands and feet seen. Symptoms were not present at birth and manifested 2 years after birth. All phalanges of hands were involved on right side and on left side, there is sparing of thumb and index finger. All phalanges of both feet were involved. There was involvement of right elbow joint causing deformity and stiffness. Other joints in body were spared. No spine abnormalities seen. Weight of patient was 24 kg and height was 117 cm, interpupillary distance was 5.6 cm, hypertelorism present, cephalic index was 79.41 (normocephalic). Forehead was prominent, straight and slightly hairy. After complete investigation, no cardiac disease, renal disease, spine disease, dental disease appreciated. Familial history of disease was present. Patient's sister who was 10 years elder than the patient was suffering from same disease and disease severity was more in sister and she died of the disease at 13 years of age. She was having acroosteolysis of distal phalanges of hands and feet, knee joint was involved, spine was involved and patient was bed ridden at age of 10 yrs, patient was not investigated for cardiac and renal disease according to attendants. Antenatal history was not significant, no developmental delay's were present at birth. Fundus examination was normal in both eyes. Audiological evaluation done, pure tone audiometry results showed bilateral moderately sensorineural hearing loss. Brainstem evoked response audiometry revealed cochlear type of sensorineural hearing loss, interpeak latency between wave I and wave III was 5.0 ms and between wave III and wave V was 9.5 ms. Impedance audiometry revealed A type graph and ipsilateral/ contralateral reflexes absent in both ears at 2000 Hz and 4000 Hz. Tone decay test was normal in both ears. In speech audiometry, SIS was 90% in left ear and 100% in right ear and SISI was 0% at 70db in both ears. Digital hearing aid trial was given to patient and patient's hearing was improved with hearing aids. Medical treatment was started by consulting orthopaedician and general physicians. Bisphosphonates, alendronate, 35 mg/week, calcium 250 mg daily prescribed to the patient.



Figure 2: Patient having deformity of all the toes.



Figure 3: Patient having deformity of all fingers of right hand and 3rd, 4th and 5th finger of left hand.



Figure 1: (A) Patient having hypertelorism, (B) Patient having stiff and deformed elbow joint.

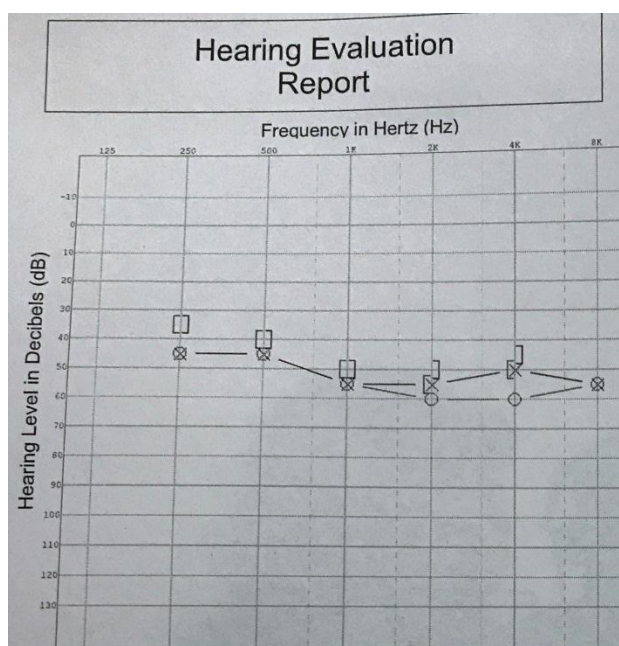


Figure 4: Pure tone audiometry showing bilateral moderate SNHL.

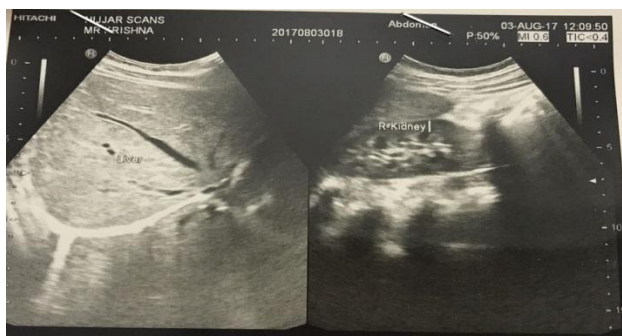


Figure 5: USG Whole abdomen.

DISCUSSION

It is a rare disease affecting any part most commonly bones. Acroosteolysis of distal phalanges, osteoporosis, skull, deformities, spinal involvement, bone fractures are common.¹⁻⁴ Heart and renal involvement may be seen.^{10,11} Hearing loss is not very common. Diagnosis is made on clinical grounds, radiological evaluation and genetic counselling. NOTCH-2 gene mutation is analysed by isolating genomic DNA from peripheral leukocytes and exon 34 amplified by polymerase chain reaction.¹⁴ Treatment is symptomatic, bisphosphonates prevent resorption of bone and hence prescribed.¹⁵ In our case, acroosteolysis of distal phalanges, hypertelorism present. No spine abnormality, heart and renal disease, dental abnormality seen. Patient presented to ENT clinic with complaints of hearing loss in both ears. Clinical and radiological evaluation and complete audiological profile done. Audiological tests revealed bilateral moderate cochlear type sensorineural hearing loss. Hearing aids and alendronate 35 mg/week prescribed.

CONCLUSION

It is a rare disorder affecting limited number of individuals. Autosomal dominant connective tissue disorder with characteristic features of acroosteolysis of distal phalanges, hypertelorism short stature, bony fractures and osteoporosis. Gene mutation NOTCH 2 on chromosome 1 is identified. Hearing loss is present in few affected individuals. Our case presented with classical history of genetic predisposition and classical features of the syndrome with complaint of bilateral moderate non-progressive hearing loss. Treatment is symptomatic. Prognosis depends on the organs affected and complications of the disease. Prognosis is not favourable in cases of neurological involvement.

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