Case Reports

Hajdu-Cheney Syndrome: First Case Report from Bangladesh

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Introduction:

Hajdu-Cheney syndrome is an extremely rare genetic disorder of the connective tissue characterized by severe and excessive bone resorption leading to osteoporosis and a wide range of other potential symtromes[1]. The clinical presentation and typical radiological features help in confirming the diagnosis[2]. Hajdu-Cheney syndrome had not been reported previously in our country. Because of the rarity of the problem, we are interested to report a case of Hajdu-Cheney syndrome who was admitted in Bangabandhu Sheikh Mujib Medical University, Dhaka, Bangladesh.

Case report:

A 15 years old girl was admitted in the paediatric ward of Bangabandhu Sheikh Mujib Medical University in February 2009 with the complaints of pain in multiple joints and progressive shortening of all the fingers of hands for 3 years. She was the 1st issue of a non-consanguineous parents and was born full-term with uneventful normal delivery. Up to 12 years of age, the girl was growing normally without any major illness. Since then she was having pain in the different joints of the hands. Pain initially started in the wrist joints and subsequently involved small joints of hands and back of the neck. There was no associated swelling or increased temperature of the joints. Movements of the fingers and hands were also normal. Later on the girl experienced severe low back pain which gradually unabled her to bend forward. She also noticed gradual shortening and widening of her fingers with some nail changes. Due to these changes she had difficulty in gripping any objects. Performing day to day household activities also became a big problem for her.

Her mother had similar illness which was so severe that she was unable to perform any routine work. Her father died 1 year back due to chest infection. She had 1 younger brother (9 years) and 1 younger sister (12 years), both of them were healthy.

On physical examination the girl had coarse facial features with antimongoloid slant, frontal bossing, mid facial flattening, slightly anteverted nostrils, long filtrum, low set ears, micrognathia and prominent occiput (Fig- 1). She also had coarse, thick hairs and short stature (height for age Z score: -- 3.5).

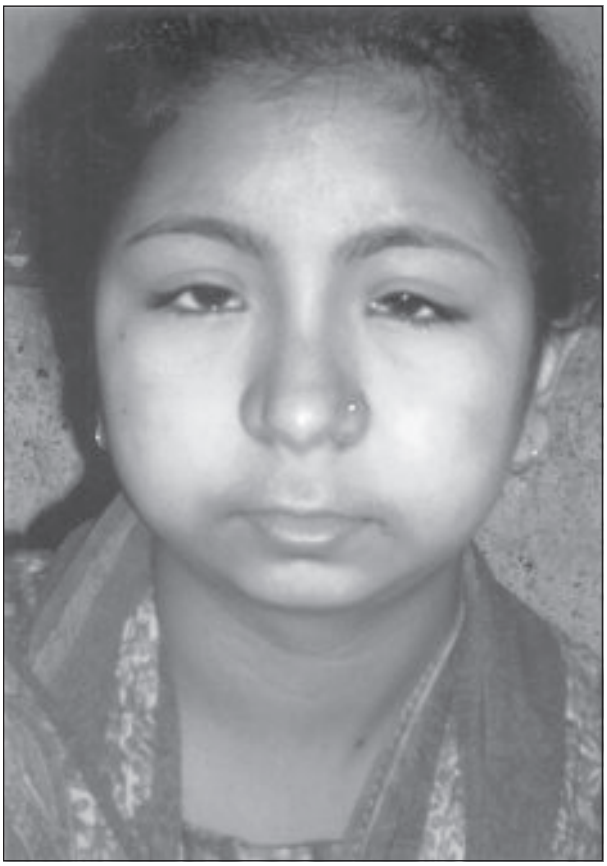

Fig.-1: *Coarse facial features including antimongoloid slant, frontal basing, long fillrum, low set ears and micrognathia.*

Her distal phalanges were shortened and clubbed (Fig-2). All the joints were hyper mobile which was documented by positive Steinberg's sign. She did not have any signs of puberty. Other examinations were normal.

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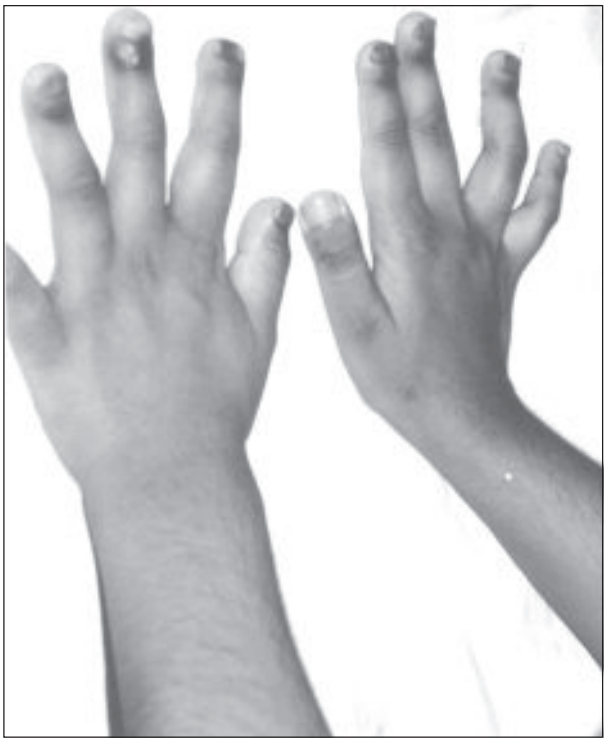

Fig 2: *Shortened and clubbed fingers.*

Routine investigations including blood count and urine was normal. The skull x-ray showed wormian bones, open skull sutures, elongated pituitary fossa and thickened skull vault (Fig – 3). X-ray hand showed gross osteopenia and resorption of the terminal phalanges with shortened triangular appearance. X-ray knee joint showed expanded epiphyseal end and gross osteopenia. X-ray para nasal sinuses showed abscence of frontal sinus (Fig – 4). From history, clinical findings and radiological evidence, the girl was diagnosed as a case of Hajdu-Cheney Syndrome. The family was counseled about the disease and the treatment options. She was prescribed calcium and vitamin D and discharged with the advice for follow up.

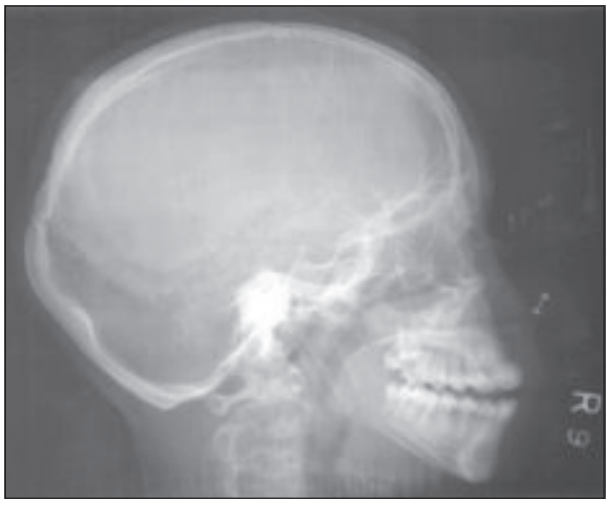

Fig 3: *Open skull sutures, wormian bones, elongated pituitary fossa and thickened skull vault.*

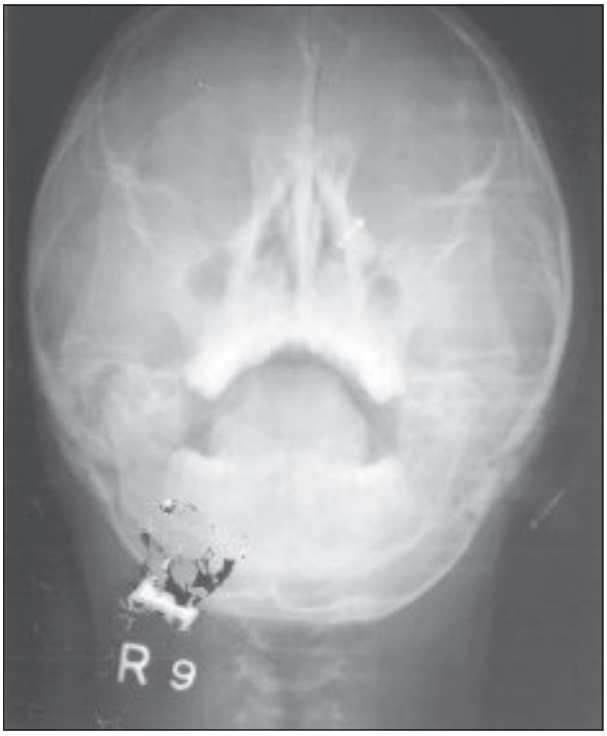

Fig 4: *Absence of frontal sinuses*

Discussion:

Hajdu-Cheney syndrome (HCS) is a rare, genetic disorder of connective tissue. Hajdu and Kauntze described this rare disorder for the first time in 1948[3]. Approximately 50 cases have been reported worldwide[4]. Among them, some were reported under the terms acro-osteolysis and arthrodento-osteodysplasia[2].

Mode of inheritance is autosomal dominant but sporadic cases are also found[2]. Though it is a congenital disorder, the correct diagnosis is rarely made until later childhood when characteristic features are found. The breakdown of bone (osteolysis), especially the outermost bones of the fingers and toes (acro-osteolysis), is a major characteristic of HCS. Clinical features of HCS also includes distinctive facial appendence, short stature, generalized, osteoporosis, joint laxity, premature loss of teeth, retarded puberty

and defective vision and hearing[5,6]. Kyphoscoliosis and basilar invagination may also be present[2].

Radiological findings include osteolysis of the distal phalanges of the hands and feet, a widened sella-turcica, thickened skull vault, generalized osteoporosis, wide cranial sutures, multiple wormian bones, hypoplasia of maxilla, mandible and absence of frontal sinuses[2]. Our patient had short stature, delayed puberty, distinctive facial appearance, joint laxity and acro-osteolysis. Radiographic findings were also consistent with HCS, which included generalized osteoponia, a widened sella turcica, presence of wormian bones, open skull sutures, acro-osteolysis, hypoplasia of maxilla, mandible and abscence of frontal sinus.

Presence of acro-osteolysis with any three other features, such as wormian bones, open skull sutures, platybasia, mid facial flattening, premature loss of teeth, coarse hair, and short stature, is enough for the diagnosis of HCS[7]. In adolescents and adults, acro-osteolysis along with family history of HCS are sufficient for the diagnosis. Most of the mentioned findings along with the family history were present in our case.

The treatment of the disorder is symptomatic. Since about 2002, some patients with HCS are given bisphosphonates to treat bone resorption associated with bone breakdown and skeletal malformations[4].

Our patient and her family was counseled regarding the disease and options for treatment. She was prescribed calcium with vitamin D. Advantages of bisphophosate was also discussed with them. But became of financial constraints they could not afford it.

Conclusion:

Hajdu-cheney syndrome is a rare hereditary disorder characterized by acro-osteolysis, skull deformities and generalized osteoporosis. Effective treatment aims to prevent bone loss, fractures and control pain. Prognosis is quite good, depending on the bone changes caused by acro-osteolysis and the neurological complications caused by basilar invagination.

References:

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