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Images in Medicine - Hajdu-Cheney Syndrome: A Rare Case Report

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A 36-year-old female patient visited the Department of Oral Medicine with the history of premature loss of multiple permanent teeth and a progressive shrinking of fingers for the past one year. Her past medical and surgical histories were not contributory. Family history revealed that she was the only child born out of a non-consanguineous marriage. She was of normal intelligence.

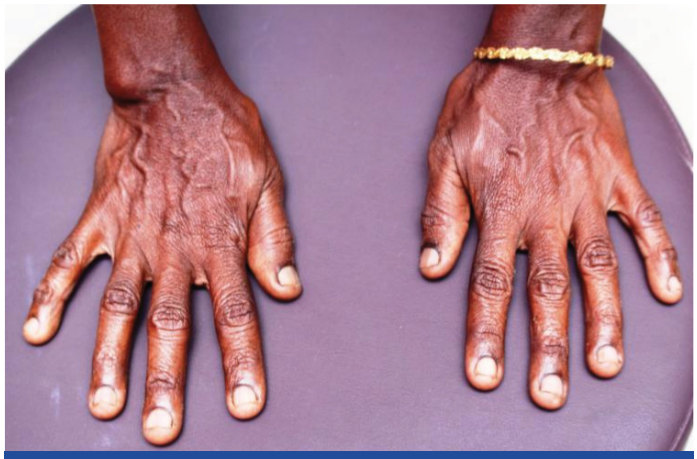
Physical examination revealed short stature, coarse hair, bushy eyebrows and prominent forehead, mid-facial flattening, slight antimongoloid slant and short neck with scoliosis. She also had prominent epicanthal and nasolabial folds with the prominent, broad nose [Table/Fig-1a&b]. She had short stubby fingers of both hands [Table/Fig-2]. Oral examination revealed multiple missing teeth with resorbed alveolar ridges of maxilla and mandible.

Laboratory investigations, which included liver function test, kidney function test, thyroid function test, parathormone levels and urinalysis, were negative. As the patient was an orphan and had no other maternal relatives, genetic analysis was not performed.

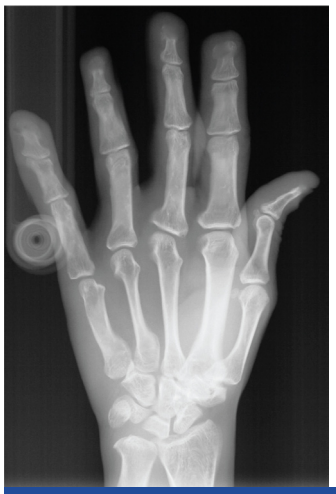
Hand wrist radiograph showed acrosteolysis of distal phalanges and subluxation at the proximal metacarpophalangeal joint of the middle finger and little finger [Table/Fig-3].

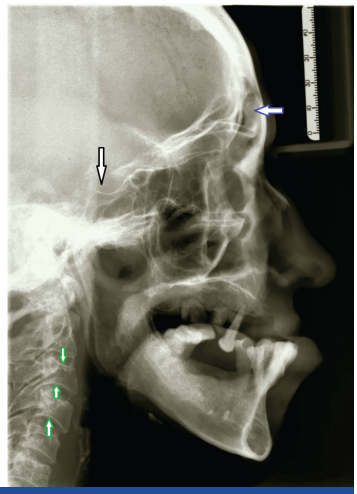
Lateral cephalogram showed an elongated 'J' shaped sella, hypoplastic frontal sinus and osteoporosis of cervical vertebrae with biconcave cervical vertebrae [Table/Fig-4].

[Table/Fig-1a & b]: Physical examination. Coarse hair, bushy eyebrows, prominent forehead, mid facial flattening, slight antimongoloid slant with prominent epicanthal and nasolabial folds



[Table/Fig-2]: Short stubby fingers of both hand





[Table/Fig-3]: Hand wrist radiograph showing acrosteolysis of distal phalanges and subluxation at the proximal metacarpophalangeal joint of middle finger and little finger
[Table/Fig-4]: Lateral cephalogram showing an elongated 'J' shaped sella, hypoplastic frontal sinus and osteoporosis of cervical vertebrae with biconcave cervical vertebrae. In this picture blue arrow indicates hypoplastic frontal sinus, black arrow indicate 'J' shaped sella and green arrows indicates biconcave cervical vertebrae

From the clinical and radiological features patient was diagnosed with Hajdu-Cheney syndrome. In most cases of HCS, treatment is based on neurological symptoms, but in this case the prognosis was good. Oral prophylaxis was done and the patient was advised to maintain good oral hygiene. To improve oral function the patient was referred to the Department of Prosthodontics for prosthodontic rehabilitation. The patient was started with oral vitamin D replacement therapy (6000 IU/day) and oral ibandronate 150 mg/month and was advised to have regular recall visits, for every three months.

DISCUSSION

Hajdu-Cheney Syndrome (HCS) is an inheritable, rare disorder of bone metabolism, associated with acro-osteolysis of the distal phalanges, premature tooth loss, and periodontitis. Main clinical features of HCS include short stature, scoliosis and kyphosis, elongation of skull, small chin, clubbing of fingers, coarse hair and thick eyebrows. Radiographically, the most frequent findings are enlarged sella turcica, wormian bones, persisting wide cranial sutures, absence of the frontal and maxillary sinuses. HCS is associated with neurologic abnormalities, such as optic nerve head swelling and mild optic neuropathy, are often the result of progressive basilar invagination [1].

Acro-osteolysis is considered to be the hallmark of syndrome which is caused by an increase in mast cells and by an elaboration of osteolytic cytokines. Molecular genetic studies have now proved that HCS is caused by a heterozygous mutation in the NOTCH gene. NOTCH is a regulator of the skeletal development and bone remodelling. Any abnormality in NOTCH signaling is

associated with developmental postnatal skeletal disorders. HCS is an autosomal dominant disorder with variable expressivity, but sporadic cases have been reported [2].

Differential diagnosis includes acro-osteolysis, premature loss of teeth, extensive osteolysis, and bizarre skull formation. HCS is of particular dental interest because craniofacial manifestations represent a constant feature of this condition. In HCS, mandible is small in childhood, may become relatively prognathic with age because of severe nasomaxillary hypoplasia [3].

REFERENCES

[1] Bazopoulou-Kyrkanidou E, Vrahopoulos TP, Eliades G, Vastardis H, Tosios K, Vrotsos IA. Vrotsos. Periodontitis associated with hajdu-cheney syndrome. *J Periodontol*. 2007;78(9):1831-38.
[2] Gupta SR, Gupta R. Hajdu-Cheney syndrome with osteomyelitis of mandible, calcification of falx cerebri and palatal groove. *The Cleft Palate-Craniofacial Journal*. 2014;51(6):722-28.
[3] Antoniades K, Kaklamanos E, Kavadia S, Hatzistilianou M, Antoniades V. Hajdu Cheney syndrome (acro-osteolysis): A case report of dental interest. *Oral Surg Med Pathol*. 2003;95(6):725–31.

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