A rare case of hypertrophic osteoarthropathy presenting as cranial entrapment neuropathy

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Introduction: Cranio- osteoarthropathy is a rare variant of hypertrophic osteoarthropathy that affect the skull , causing the increased bone growth and decreased ossification, leading to compression of cranial nerves while they pass through narrow channels, resulting in cranial entrapment neuropathy.

Aim :

To describe the clinical aspects of a 8 year old male with bilateral facial paresis with sensorineural hearing loss.

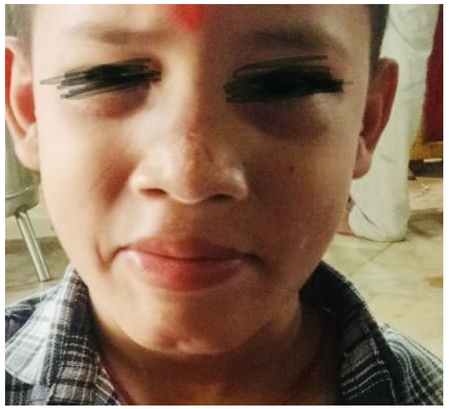
Case report :

A 8 year-old male, born out of 3rd degree consanguineous marriage with normal birth and developmental history. History of present illness started at 4 years of age as deviation of angle of mouth to left , associated with drooling from right corner of mouth and difficulty to close right eye completely. After 2 months, the child was unable to close the mouth a with drooling from both the corners of the mouth and eyes left half open during sleep, unable to smile, with slurring of speech. After 6 months , he developed hardness of hearing , more in the right compared to left ear. Able to hear to some extent only when spoken loud close to the ear. On examination, there was **bilateral lower motor neuron type facial paresis** with **bilateral sensorineural hearing loss**. Other cranial nerves, motor, sensory and cerebellar examination were unremarkable. No history of similar complaints in family.

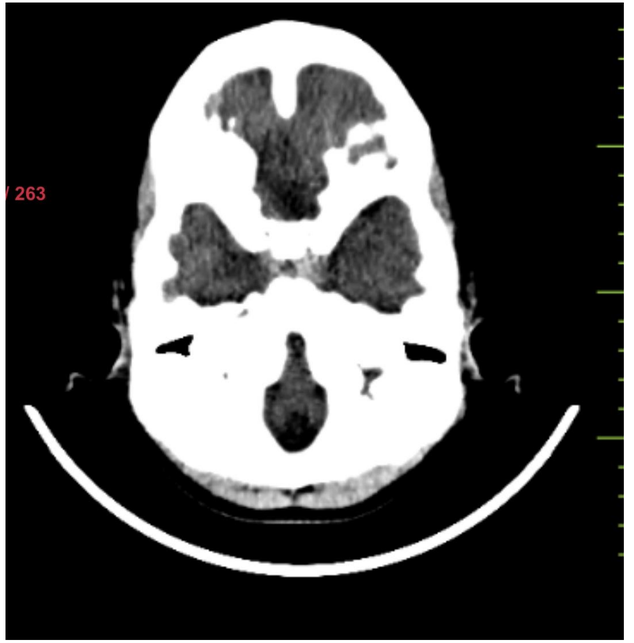
Results :

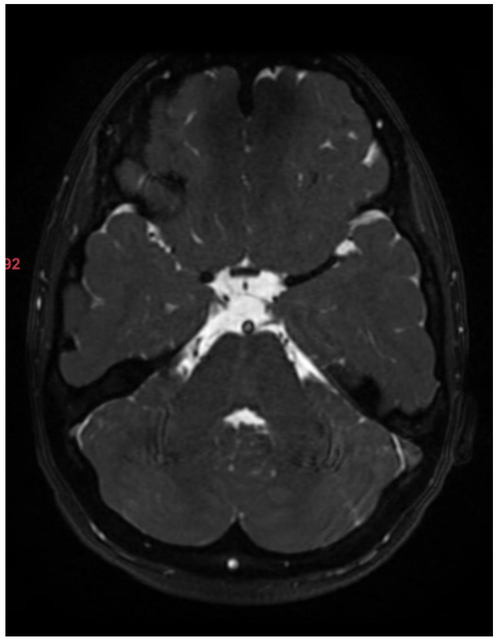
S. Calcium -9.5mg%, s. Phos- 5.2mg%, ALP-=683,
Skeletal survey showed increased cortical thickness and density in skull bones base with loss of normal diploic spaces between skull bones. Long bones, ribs, vertebra, wrist and feet bones showed mild increase in cortical density without any fractures.

CEMRI brain with 3D FIESTA showed narrowing of bilateral internal acoustic meatus (right - 1.8mm , left - 1,7mm) with compression over bilateral vestibulocochlear and facial nerves. **BERA** -right>left sensorineural hearing loss, **Blink** - bilateral facial not recordable.
Whole exome sequencing showed SLCO2A1 gene mutation in heterozygous state .



Gene, Variant details	Zygosity Depth (Alt allele %)	In silico tools	MAF	Literature	OMIM Disease	Inheritance
SLCO2A1 **(-)** c.296G>A p.Arg99His ENST00000310926.11	Heterozygous 132X(47.7%)	SIFT - D LRT - N PolyPhen - PrD MT2 - D	1000 G - NA gnomAD (V2.1) - 0.01% gnomAD (V3.1) - 0.006% MedVar - 0.03%	-	Hypertrophic osteoarthropathy, primary, autosomal dominant (OMIM#167100)	Autosomal dominant





Discussion:

Cranio-osteoarthropathy is a rare form of primary hypertrophic osteoarthropathy (PHO) also known as Touraine- Solente- Gole syndrome, characterized by abnormal bone remodeling, particularly affecting the skull base. The SLCO2A1 gene mutation identified in this patient leads to impaired prostaglandin E2 (PGE2) transport, resulting in elevated PGE2 levels, which promote periosteal new bone formation and cortical thickening. While PHO typically presents with digital clubbing, periostosis, and joint symptoms, **cranial nerve involvement is uncommon and represents a rare and severe manifestation of skull base sclerosis**. The constellation of **elevated ALP**, **increased bone density**, and **cranial neuropathies** should raise suspicion for an underlying **genetic bone dysplasia**. Early recognition and diagnosis are essential to guide management, monitor for progressive neurological compromise, and offer genetic counseling.

Conclusion:

In our case, the bone density in the skull base is affected , for which no definite treatment is currently available to halt the progression. Surgical decompression of cranial nerves can be performed to relieve pressure and prevent further potential damage. Further studies are needed to understand the phenotypic spectrum associated with heterozygous SLCO2A1 mutations and their implications in bone metabolism and cranial neuropathies.