

Craniofacial dysostosis

General information

- Inherited as an autosomal dominant trait
- in some cases affected individuals have no family history of disease
- disorders characterised by distinctive malformations of the skull and facial region

Clinical features

- It is a rare genetic disorder that may be evident at birth or during infancy
- the fibrous joints between certain bones of the skull close prematurely
- facial abnormalities typically include unusual bulging or protrusion of the eyeballs due to shallow eye cavities
- premature closing of suture may result in brachycephalic that is short head
- in extreme cases patient may demonstrate Clover leaf skull

Oral manifestations

- Maxillary hypoplasia which shortened anteroposterior dimensions of maxillary arch
- facial angle is exaggerated and the patient nose is prominent and pointed resembling parrot beak
- dental arch which is reduced and this gives an appearance of high arched palate
- unilateral or bilateral crossbite is evident with open bite and crowding in mandibular teeth. Shovel shaped maxillary incisors cleft lip and palate are also evident

Radiographic features

- Digital marking in skull as a result of increased intracranial pressure from early synostosis of cranial sutures
- the cranial walls are thin with multiple radiolucencies appearing as depressions or scalloped appearance of beaten silver
- malformation of calvarium, flattened mandibular angle, conical teeth and partial anodontia

Diagnosis

- High arched palate, with midface deformity, proptosis is typical feature of Crouzon syndrome
- beaten silver appearance seen in rows and syndrome

Management

- Midfacial advancement
- craniectomy – it is needed to alleviate the raise intracranial pressure
- fronto orbital advancement – this is done to correct ocular defect