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Case Report

RAINE SYNDROME: a rare case presentation

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Abstract: Raine syndrome is a rare, lethal,autosomal recessecive disorder with characteristic features of exophthalmos, choanal atresia or stenosis, osteosclerosis and cerebral calcifications. Most of babies with this disorder die shortly within few weeks after birth. We report a baby who was 3 days old at the time of presentation.

Keywords: Lethal, osteosclerosis, Raine syndrome

Introduction:

Raine syndrome is a neonatal osteosclerotic bone dysplasia of early and aggressive onset that usually results in death within the first few weeks of life, although there have been some reports of survival into childhood. Radiographic studies show a generalized increase in the density of all bones and a marked increase in the ossification of the skull. The increased ossification of the basal structures of the skull and facial bones underlies the characteristic facial features, which include narrow prominent forehead, proptosis, depressed nasal bridge, and midface hypoplasia. Periosteal bone formation is also characteristic and differentiates it from osteopetrosis and osteosclerotic bone dysplasias, typically extends along the diaphysis of long bones adjacent to areas of cellular soft tissue. Extra-skeletal features can be hydronephrosis and ureteral stenosis.

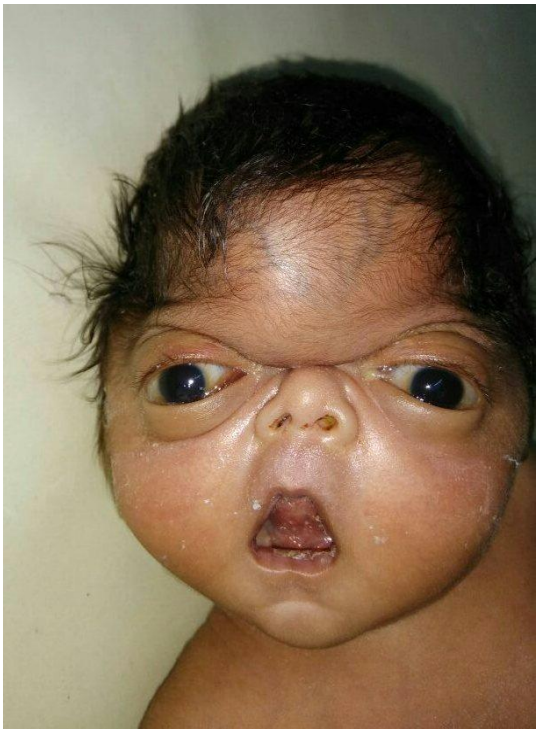
Case report:

This 3 day old female chid born to non-consanguineous parents, presented at opd with history of increase in respiratory effort and difficulty in feeding since 2nd day of life. Initial evaluation suggestive possibility of upper airway obstruction as the child was having persistent open mouth breathing with facial feature of depressed nasal bone and failed attempt of passing infant feeding tube through both nostrils. Child admitted in neonatal care unit and further care taken to establish airway, breathing and circulation. Infantogram done suggestive of generalised osteosclerosis. Oxygen supplemented & IVF started , general condition improved within 48 hrs.On detailed physical examination, child was having large open anterior fontanel (10*11cm) and posterior fontanel(3*3cm), bilateral exopthalmos with corneal dryness, depressed nasal bridge, long philtrum, large frontal encephalocele, low set ears, prominent scalp vein ,high arched palate. On systemic examination IFT could not be passed with no apparent lower respiratory tract abnormality noted, spine and back were also normal, CVS finding was grade 2 systolic murmur, CNS examination were normal ,abdomen were mildly distended no apparent organomegaly noted,

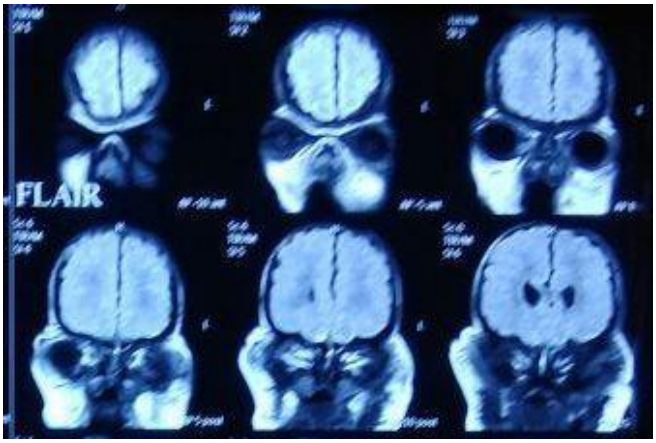
Routine investigation like CBC, RFT were normal. Infantogram showing osteosclerosis of bone, MRI suggestive of cloverleaf shaped skull with focal areas of skull vault thining/nonvisualisation, wide open fontanel and sutures noted through which bulging of brain tissue noted. medial bowing of lateral wall of nasal cavity on either side with narrowing of choanal airspace with thickening of vomer bone suggestive of bilateral bony choanal atresia, multiple foci of internal calcification at bilateral periventricular and subependymal location, bilateral basal ganglia, left posterior parietal white matter ,splenium of corpus callosum and bilateral tentorium cerebelli. Hypoplastic bony orbital wall with shallow orbit with protosis, hypoplastic nasal maxilla pterigoyid, palatal bone, coronal and condylar process of maxilla. Mild prominence of parietal and temporal horn of left ventricle noted. Non visualisation of pituitary gland. 2D echo small PFO,USG ABD-KUB was normal.

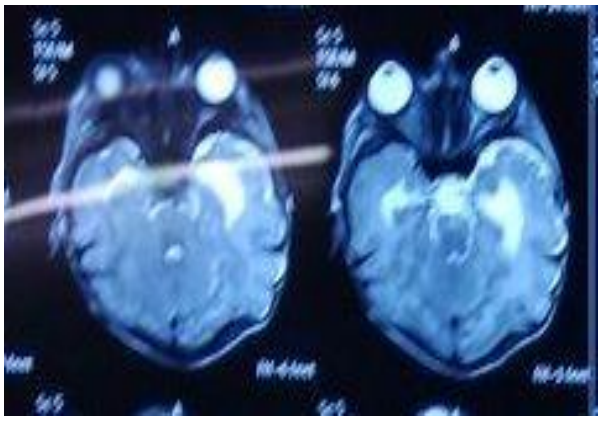
Disscusion:

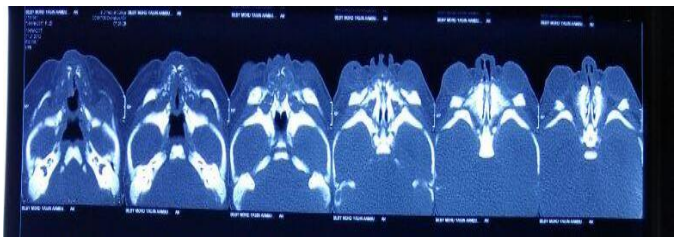
Raine syndrome, also known as lethal osteosclerotic bone dysplasia, is a rare autosomal recessive disorder characterized by exophthalmos, microcephaly, depressed nasal bridge, bilateral choanal atresia/stenosis, gum hyperplasia and osteosclerosis.The osteosclerosis is usually generalized, but occasionally may be focal involving only few bones. Palate usually shows cleft, but may be high arched and narrow. The babies affected with this disorder show intracranial calcifications.Calcifications are seen in parietal and occipital periventricular white matter and corpus callosum, but never occur in cortex, temporal lobes, internal capsule, cerebellum and brain stem.

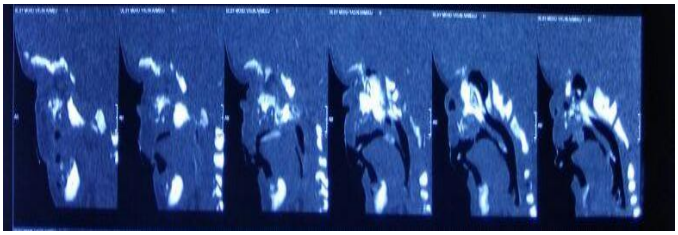


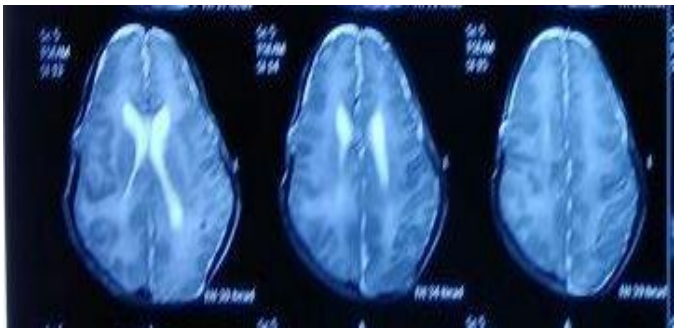
Major association between sclerosing dysplasia and intracranial calcification is seen in osteopetrosis associated with renal tubular acidosis and carbonic anhydrase II deficiency .However, in this disorder, the calcifications are usually seen after 2 years of age exclusively in basal ganglia and cortex and characteristic clinical features of Raine syndrome are lacking.











It is inherited as autosomal recessive; mutations in FAM20C gene on chromosome 7 are identified in babies with this disorder.

Initial reports showed that this disorder is lethal and most of babies with this disorder died in early months of their life. However, mutations in FAM20C gene were also noticed in older children with phenotypic features of Raine syndrome

with osteosclerosis, indicating that lethality is not essential for diagnosis.

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