

Raine Syndrome: A Case Report

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Abstract: Raine syndrome, also known as lethal osteosclerotic bone dysplasia, is a rare genetic disorder with characteristic features of exophthalmos, microcephaly, depressed nasal bridge, bilateral choanal atresia, gum hyperplasia, osteosclerosis & cerebral calcification. Most of babies with this disorder die immediately after birth. We report a case of baby who is 2 days old at the time of presentation.

Keywords: Raine Hereditary Osteosclerosis Microcephaly

Case Report

A 2 days old baby, born to consanguineous parents, was referred to Civil Hospital Ahmedabad for multiple congenital anomalies like absent or hypoplastic nasal bone, bilateral ocular proptosis, craniosynostosis & skeletal dysplasia on antenatal USG. (Figure I) The baby cried soon after birth. This was the third baby born to parents. The first two children are normal. Physical examination was significant for multiple congenital malformations including microcephaly, bulging wide fontanelles, narrow prominent forehead, bilateral proptosis, low-set ears, mid-face hypoplasia, hypoplastic nose with bilateral choanal atresia, and micrognathia/retrognathia. (Figure I)

Physical examination revealed stable vitals & there was no cyanosis. Baby was weighing 3 kg. Routine investigations were done & the baby was admitted for further evaluation. Baby was fed with expressed breast milk. There was small nose with depressed nasal bridge, high arched narrow palate & long philtrum. Infant feeding tube could not be passed through the nose indicating associated choanal atresia/stenosis. Baby's lungs were clear, central nervous system & abdominal examinations were normal. Subsequently, investigations like USG – Abdomen-Pelvis & Brain, X-rays – Infantogram, 2-D echo, CT scan of Brain & PNS & MRI of brain were done.

Baby's routine blood investigations were normal. Serology for TORCH infection was negative. Infantogram of baby showed generalised osteosclerosis. X-Ray skull showed increased bone density. (Figure II A) The medullary cavities of long bones were poorly differentiated from cortex & irregular periosteal thickening were present. (Figure II B). USG of brain showed abnormal periventricular hyperechogenicity. CT scan of brain showed calcified lesions in bilateral periventricular & subependymal locations, bilateral basal ganglia, splenium of corpus callosum & along tentorium cerebelli. (Figure III A). CT scan of PNS showed bilateral bony choanal atresia with mid face hypoplasia. (Figure III B) MRI of brain showed wide open fontanelles & sutures through which bulging of brain tissue noted with resultant frontal bossing. (Figure IV A). Multiple signal void intensities are noted along bilateral periventricular & subependymal locations, bilateral basal ganglia, splenium of corpus callosum & along tentorium cerebelli, correlating with CT findings, suggestive of intracranial calcifications. (Figure IV B) Mild prominence of parietal (9 mm) & temporal horn (10 mm) of left lateral ventricle is noted. (Figure IV B). Then patient advised for genetic testing. Unfortunately, the baby died before the genetic testing was done. Diagnosis of Raine's syndrome was made based on typical clinical presentation & radiographic findings.

Figures

Figure I



Figure II A



Figure II B



Figure III A

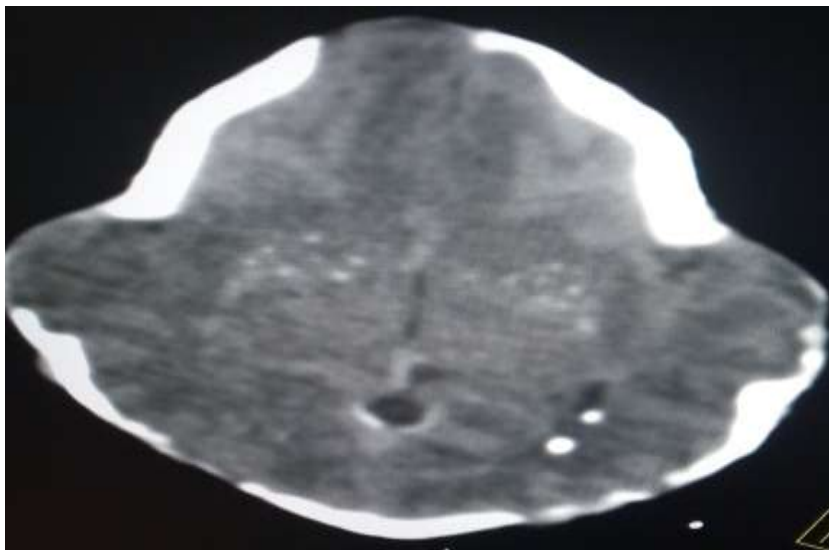


Figure III B



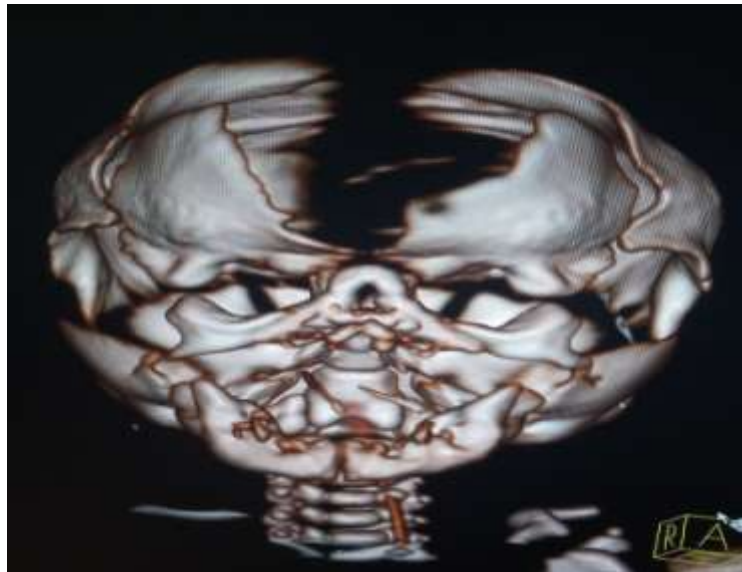
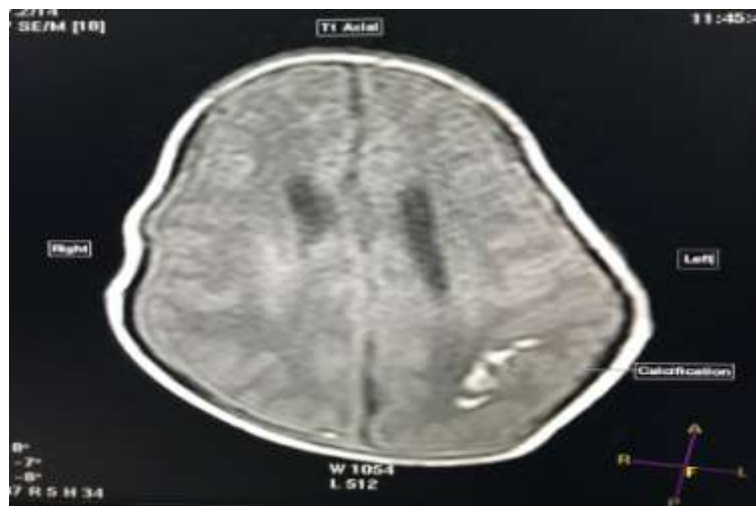


Figure IV A



Figure IV B



DISCUSSION

Raine syndrome, also known as lethal osteosclerotic bone dysplasia, is a rare autosomal recessive disorder characterised by exophthalmos, microcephaly, depressed nasal bridge, bilateral choanal atresia, gum hyperplasia, osteosclerosis & cerebral calcification. It is first described by Raine et al' in 1989(1) It is inherited as autosomal recessive; mutations in FAM20C gene on chromosome 7 are identified in babies with this disorder. Originally, Raine syndrome was reported as a lethal syndrome. However, recently a milder phenotype, compatible with life has been described. Biallelic variants in FAM20C, encoding a Golgi casein kinase involved in bio mineralisation, have been identified in affected individuals.(2) The osteosclerosis is usually generalised, but occasionally may be focal involving only few bones. Palate usually shows cleft, but may be high arched & narrow. Cleft of mandible & maxillae have been described by Kan & Kozłowski(3) & the associations with posterior encephalocele have been described by Al Mane et al(4). Abnormalities in urogenital tract have been described in patients with Raine's syndrome, including bilateral hydronephrosis, & stenotocostia of both ureters. Pulmonary hypoplasia is common associated feature. Cardiomegaly with left ventricular muscular hypertrophy & hypoplasia of the main stem pulmonary arteries have been reported in association with Raine's syndrome.

The babies affected with this syndrome show intracranial calcifications. Calcifications are seen in parietal & occipital periventricular white matter & corpus callosum. Major association between sclerosing dysplasia & intracranial calcification seen in osteopetrosis associated with renal tubular acidosis & carbonic anhydrase II deficiency.(5) However, in this disorder, the calcification are usually seen after 2 years of age exclusively in basal ganglia & cortex and characteristic clinical feature of Raine syndrome are lacking.

References

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