CASE REPORT

FAMILIAL CROUZON'S SYNDROME WITH PATENT DUCTUS ARTERIOSUS: A RARE COINCIDENCE

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ABSTRACT: Recently we operated a case of large patent ductus arteriosus (PDA). On clinical examination and radiological investigation patient has features of rare Crouzon's syndrome. Familial affection are usually mother and her daughter but in this case father and not mother affected. Being a rare anomaly and has association with cardiac anomaly and familial tendency, we feel it is worth to report this patient.

KEYWORDS: Patent ductus arteriosus, Crouzon's syndrome, Branchial arch syndrome, Genetic disorder.

INTRODUCTION: Crouzon's syndrome is a developmental anomaly of first branchial arch. It is a rare anomaly affecting mainly skull face with prognathism, low set ear with hearing loss, high arch palate and cardiac anomalies mainly PDA.^(1,2,3) It is an autosomal dominant with genetic mutation and may have familial tendency.^(3,4,5,6,7)

CASE REPORT: A 9 year old female child with body weight of 14 kg and height of 105 cm presented with repeated respiratory tract infection and failure to thrive. On general examination she has broad forehead prominent bulging eyes, prognathia, low lying external ears, high arch palate deformity, mental retardation, deafness and growth retardation with associated kyphoscoliosis.

FAMILY HISTORY: Father has similar facial deformity but no other anomalies. Cardiac examination shows apex at 6th intercostal space mid clavicular line. S1 normal P2 loud, There was continuous machinery murmur infraclavicular and pulmonary region. ECG Normal sinus rhythm, normal axis, p mitral, Left Ventricular (LV) hypertrophy, incomplete LBBB. X ray chest- Kyphoscoliosis deformity present CTR>50% LV apex, Aortic Knuckle prominent Pulmonary conus prominent pulmonary plethora present. 2D Echo- Large patent ductus arteriosus with dilated Left Atrium, LV normal function.

OPERATIVE DETAIL: Patient was taken for PDA closure. PDA double ligation was performed via left posterolateral thoracotomy approach. PDA was dissected retro pleurally, it was large and of size of descending aorta. Under hypotensive anaesthesia it was looped twice and ligated. Drain kept, chest closed in layers. Patient extubated shifted to ICU and did well postoperatively.

DISCUSSION: This congenital anomaly was named after Octave Crouzon,⁽⁸⁾ a French physician. It is caused by mutation in fibroblast growth factor receptor II located on chromosome 10.^(3,4,5,6,7) Incidence of crouzon's syndrome is currently estimated to occur in 1.6 out of every 1,00,000 people.⁽⁶⁾ There is a great frequency in families with a history of the disorder, but that does not mean that

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everyone in the family is affected. Diagnosis can occur with sign and symptom, clinical examination, radiograph, magnetic resonance imaging scan, genetic testing, CT scan and diagnosis of associated anomalies like cardiac with 2D Echo. It is an autosomal dominant genetic disorder⁽⁸⁾ known as a branchial arch syndrome affect first branchial arch mainly affecting maxilla and mandible.^(1,7) Low set ears, ear canal malformation with hearing loss and in severe cases Meniere's disease may occur.

Craniosynostosis^(2,9) with short and broad head, Exophthalmos, hypertelorism (greater than normal distance between eyes), Psittichorhina (beak like nose), Hypoplastic maxilla (insufficient growth of mid face) result in prognathism (chin appears to protrude despite normal growth of mandible).^(2,8,9) It is associated with PDA and aortic coarctation.⁽⁹⁾ Other anomalies are shorter humerus and femur to rest of body. A small percentage has partial syndactyly. Oral and maxillofacial, dental, plastic, and associated cardiac surgery help patient.^(1,2,3)

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Fig. 1: Abnormal denture and High arched palate

Fig. 2: Hypertelorism, Low set ears, broad Fore head

Fig. 3: Postoperative scar of PDA closure

Fig. 4: The patient with her father

Fig. 5: X-RAY skull AP and Lateral

Fig. 6: X-RAY Chest PA view

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