|| ISSN(online): 2589-8698 || ISSN(print): 2589-868X ||

Available online on http://www.ijmbs.info

International Journal of Medical and Biomedical Studies; 2025; 9(5); 63-68

Case Report

Arteriovenous Malformation in a Patient with Crouzon Syndrome

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Received: 02-10-2025 / Revised: 24-10-2025 / Accepted: 16-11-2025
DOI: https://doi.org/10.32553/ijmbs.v9i5.3124
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Conflict of interest: No conflict of interest

Abstract:

Introduction: Crouzon syndrome is a rare autosomal dominant genetic disorder associated with a mutation in the fibroblast growth factor receptor 2 (FGFR2) gene, which affects the first branchial arch. It is characterised by premature craniofacial dysostosis, resulting in skull deformities, facial anomalies, and proptosis.

Patient Presentation: We report a case of Crouzon syndrome who presented with no neurological symptoms and was incidentally found to have arteriovenous malformations in the vertex and occipital region.

Management and outcome: This case elucidates the importance of considering vascular anomalies in patients with syndromic craniosynostosis, and imaging strongly contributes to early identification of AVMS and can guide multidisciplinary evaluation and intervention to optimise outcomes in such complex presentations.

Conclusion and contribution: The association between Crouzon syndrome and AVMs remains poorly understood; therefore, further studies and reporting of similar cases may help establish a better pathophysiological link.

Keywords: Crouzon, Craniosynostosis, Arteriovenous malformations

Introduction

Crouzon syndrome is a rare genetic disorder resulting from mutations in the fibroblast growth factor receptor 2 (FGFR2) gene on chromosome 10q25-26. It is characterised by craniosynostosis with resultant craniofacial and dental anomalies. The skeletal manifestations and the vascular anomalies, such as AVMs, are well documented in the literature. As these AVMS are high flow connections between arteries and veins carry the risk of haemorrhage, seizures and neurological deficits, having the potential to alter the clinical course and therapeutic approach. This case not only adds to the literature on vascular anomalies like AVM in Crouzon syndrome but also other craniosynostosis syndromes, and lays emphasis on the need

for heightened clinical suspicion and imaging evaluation.

Patient Presentation

A male child in his early years was brought to the paediatric outpatient department of the tertiary care hospital by his parents with concerns regarding delayed developmental milestones. The parents reported that the child had been treated by a local paediatrician without any definite diagnosis for not achieving age-appropriate speech and motor skills, and was under his/her follow-up. There was no history of seizures, vision disturbances or limb weakness. The antenatal history was unremarkable, and there were also no perinatal complications. Family history was non-contributory as there were no known hereditary disorders or similar complaints in siblings or close relatives.

Management and Outcome

On physical examination, the child has distinctive craniofacial features with a prominent forehead, exophthalmos, midface hypoplasia and a beaked nasal bridge. The cranial shape was abnormal, and two palpable defects were noted in the vertex and occipital regions of the skull, with soft, warm pulsating lesions with palpable thrill felt over the defects. The neurological physical examination revealed hypotonia; however, no focal neurological deficit was seen.

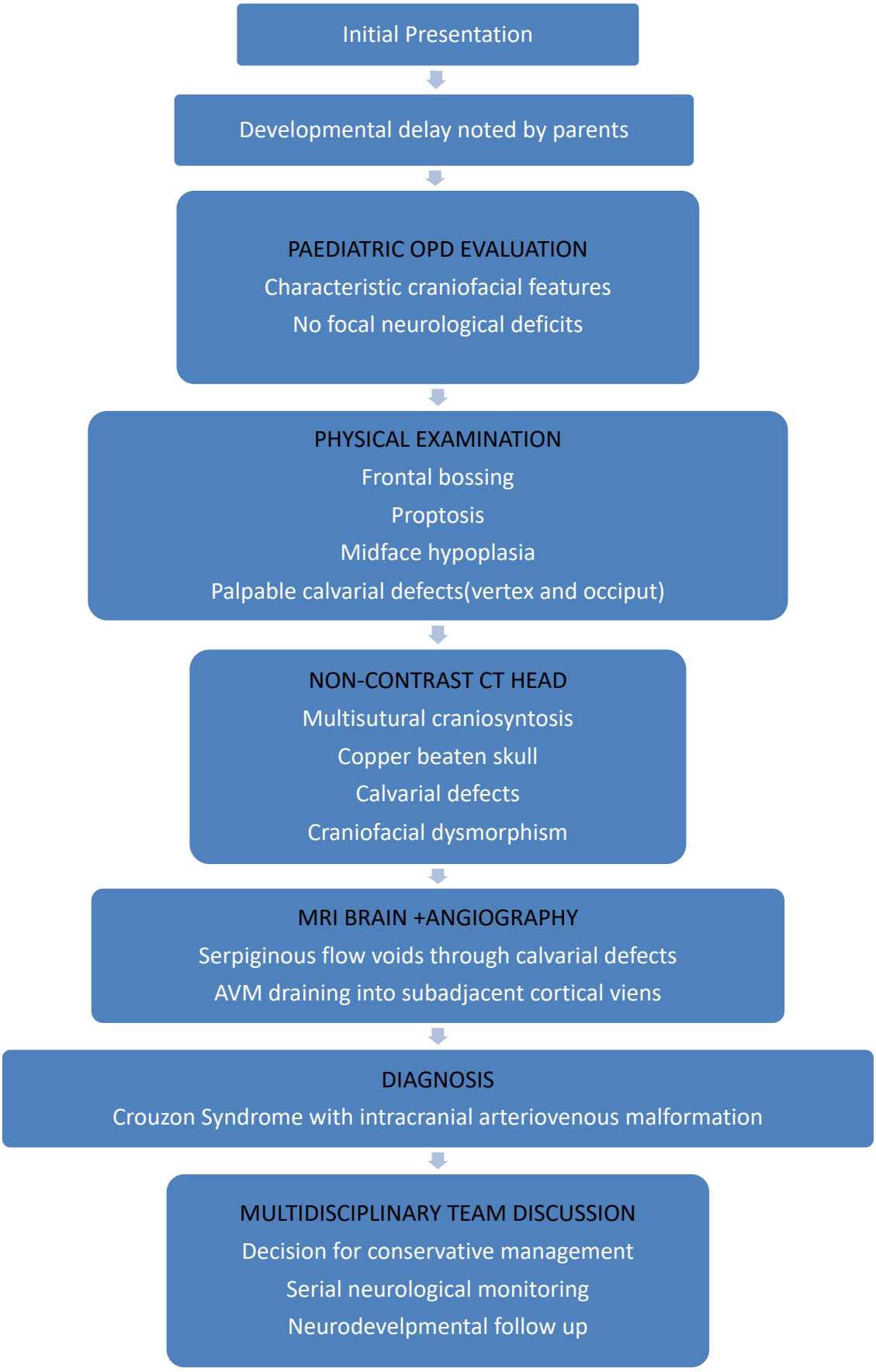
The baseline laboratory investigations, including metabolic and endocrine profiles, were within normal limits.

Non-contrast Computed tomography (NCCT) of the head demonstrated multi-sutural craniosynostosis, with premature fusion of the coronal, sagittal and lambdoid sutures. There is of prominent protruding forehead, and the calvaria showed copper copper-beaten appearance, suggesting raised intracranial pressure. The maxilla and zygomatic bones are underdeveloped with resulting concave facial profile. There was ocular protrusion due to shallow orbits, as there is reduced anteroposterior orbital depth. The paranasal sinuses are hypoplastic. Calvarial defects are seen in the midline at the vertex and occipital region.

Subsequently, Magnetic resonance imaging (MRI) of the brain and face was performed, which further delineated the craniofacial abnormalities and confirmed the presence of two midline calvarial defects in the vertex and occipital region. These defects showed the presence of serpiginous flow voids evident on T1 and T2 weighted images, indicating high-flow vascular malformations. The contrast-enhanced MR angiography was done, which revealed the presence of a nidus of arteriovenous channels coursing through these defects and draining into the subjacent cortical veins. No haemorrhage or hydrocephalus was evident on both CT and MRI. The clinical as well as imaging findings supported the diagnosis of Crouzon syndrome with associated intracranial arteriovenous malformation**(Illustration 1).**

Table 1: Differential Diagnosis

Type	Crouzon Syndrome	Apert Syndrome	Pfeiffer Syndrome
Pattern of Craniosynostosis	Coronal with or without sagittal/lambdoid, multi-sutural	coronal	coronal
face	Proptosis	High, prominent forehead	Proptosis
Limbs	Not involved	Cutaneous syndactyly	Wide thumb and toe
Intellectual disability	Usually normal cognition unless secondary complication	May be impaired	Impaired only in severe case
Other systemic features	No major visceral anomalies	Possible cardiac, genitourinary anomalies	Possible tracheal cartilage anomalies


Initial Presentation
Developmental delay noted by parents
PAEDIATRIC OPD EVALUATION
Characteristic craniofacial features
No focal neurological deficits
PHYSICAL EXAMINATION
Frontal bossing
Proptosis
Midface hypoplasia
Palpable calvarial defects(vertex and occiput)
NON-CONTRAST CT HEAD
Multisutural craniosyntosis
Copper beaten skull
Calvarial defects
Craniofacial dysmorphism
MRI BRAIN +ANGIOGRAPHY
Serpiginous flow voids through calvarial defects
AVM draining into subadjacent cortical viens
DIAGNOSIS
Crouzon Syndrome with intracranial arteriovenous malformation
MULTIDISCIPLINARY TEAM DISCUSSION
Decision for conservative management
Serial neurological monitoring
Neurodevelpmental follow up

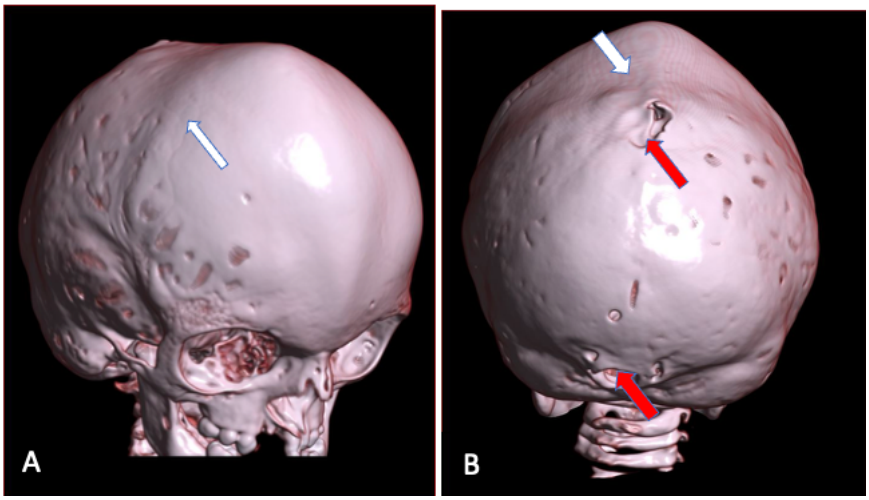


Figure 1: 3D images Volume rendered images of the young child of Crouzon syndrome (A and B) shows presence multiple sutural cranio synostosis i.e. coronal, sagittal and lambdoid (white arrows). There is presence of midline cranial defects present in vertex and occipital region (red arrows)

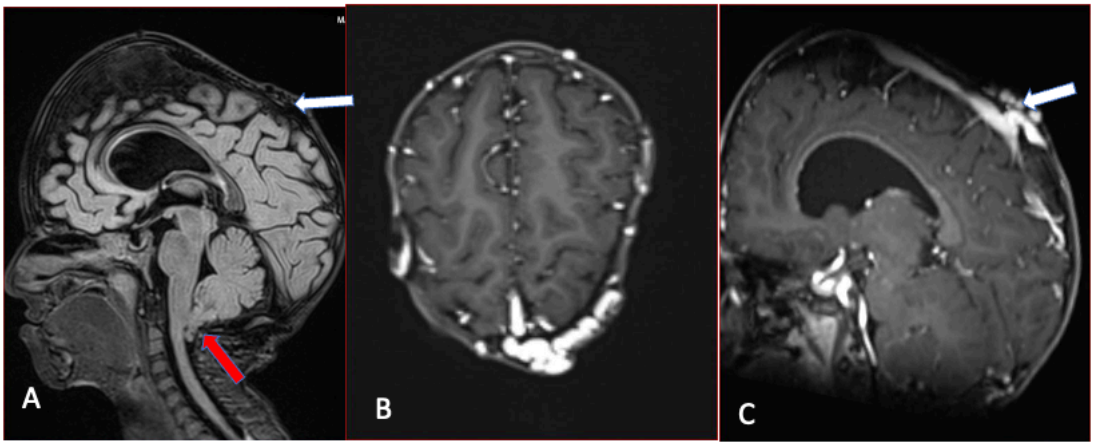


Figure 2: The MRI images Sagittal and axial (A-FLAIR, B,C-post contrast T1 weighted) shows tuft od serpiginous structures seen in vertex seen as flow voids in Image A(arrow) and with enhancement in image C. There is presence of Chiari 1 malformation seen (red arrow in image A)

Discussion

Crouzon syndrome is a rare genetically inherited disorder with an incidence of 1.6:1,00,000 live births resulting from mutations in the fibroblast growth factor receptor 2 (FGFR2) gene on chromosome 10q25-26 [1,2]. It was first described in 1912 by Louis Edouard Octave Crouzon, a French neurologist and geneticist [3]. The CT scan images show characteristic features of premature craniosynostosis of multiple sutures and consequent skull and facial deformities, which often manifest as a triad of skull shape abnormalities, facial deformities and exophthalmos [1,2], which was seen in our case. The resultant deformities vary due to the different sutural involvement, either single or in combination. Sutural synostosis could be brachycephaly, dolichocephaly or trigonocephaly [8]. The ophthalmic manifestations too are variable and can be proptosis, hypertelorism and visual acuity abnormalities [9]. Our case shows a nose beak face having retrusion, a jaw showing prognathism and a mouth showing overcrowding of teeth with a high arched palate [10] along with the premature craniosynostosis with fusion of coronal,

lambdoid and sagittal sutures. Two defects were seen in the midline calvaria in the vertex and occipital region, showing the presence of arteriovenous malformations draining into sub-adjacent cortical veins, which were evident on MRI [4]. The probable pathophysiology for the occurrence of AVM is likely to be multifactorial, possibly due to craniosynostosis altering venous hemodynamics and structural bone defects acting as conduits for aberrant vessels. Literature describes associations of Crouzon syndrome with Chiari 1 malformation, which may be present in 70% of cases. Hydrocephalus, agenesis of corpus callosum and cervical spine fusion may also be associated in a few cases [4,5]. The associated findings in our case were the presence of Chiari 1 malformation and hydrocephalus. These findings or complications, though asymptomatic at present but are liable to progress into severe life-threatening conditions[6] (**Illustration 2**). In non-life-threatening conditions, the combined approach of surgical, medical and supportive interventions is needed for management of functional impairments and improving the quality of life [7].

Conclusion and contribution

Thus, Crouzon syndrome, having characteristic features of craniosynostosis along with craniofacial manifestations, can be associated with intracranial arteriovenous malformations, and timely recognition with appropriate radiological imaging is necessary as these AVMs can lead to serious neurological complications.

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