

## Bilateral squamosal suture synostosis: A rare form of isolated craniosynostosis in Crouzon syndrome

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**Core tip:** We report the unique case of isolated bilateral squamosal suture synostosis in a 13-mo old patient with Crouzon syndrome. Although bicoronal sutures are most commonly involved in patients with Crouzon syndrome, it is important to be familiar with this rare entity of craniosynostosis in order to prevent misdiagnosis or delayed surgical treatment.

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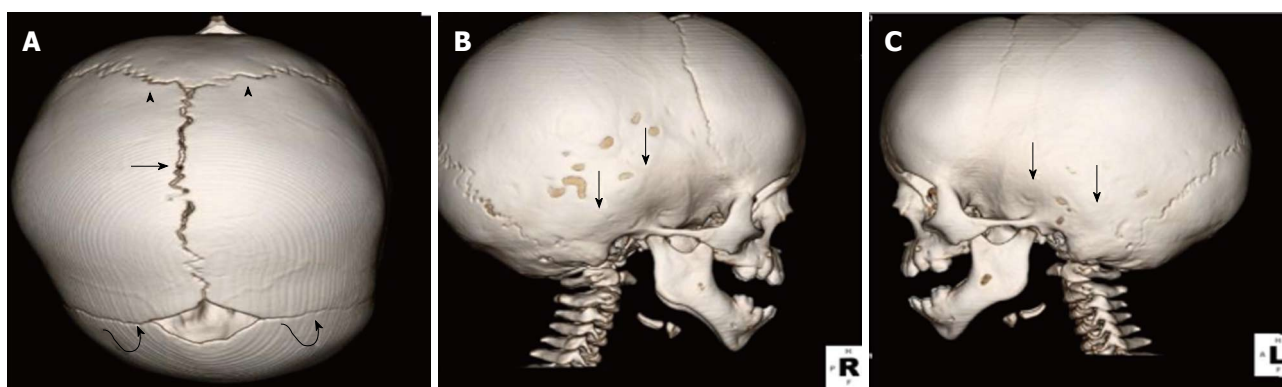
### Abstract

Craniosynostosis is a pathologic condition which is characterized by the premature fusion of cranial sutures. It may occur alone or in association with other anomalies making up various syndromes. Crouzon syndrome is the most common craniosynostosis syndrome. Bicoronal sutures fusion is most commonly involved in Crouzon syndrome. There have only been a handful of cases of squamosal suture synostosis described in the surgery literature with the few ones described in Crouzon syndrome associated with other types of craniosynostosis. To the best of our knowledge, we are presenting the first case of isolated bilateral squamosal suture synostosis in a patient with Crouzon syndrome in a radiology journal with emphasis on its radiological appearance.

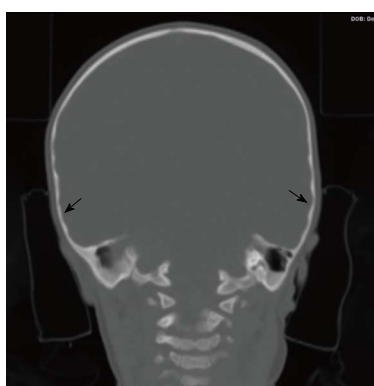
### INTRODUCTION

Crouzon syndrome is a very rare autosomal dominant craniofacial dysostosis that occurs in approximately 16 in 1 million live births<sup>[1-3]</sup>. It was described in 1912 by a French neurologist, Octave Crouzon, in a mother and son exhibiting a triad of skull deformities, facial anomalies and exophthalmos<sup>[4]</sup>. Typically Crouzon syndrome has been associated with bicoronal synostosis leading to a characteristic tall, flattened forehead. Midface hypoplasia, a beaked nose, and proptosis have also been described as characteristic features<sup>[5,6]</sup>.

To the best of our knowledge, there have been no cases of Crouzon syndrome associated with isolated bilateral squamosal suture craniosynostosis described in the literature. We report the radiological appearance of bilateral premature squamosal craniosynostosis found in a 13 mo-old female with Crouzon syndrome during her work



**Figure 1** 3 D computed tomographic image. A: Of the skull demonstrating normal appearance to the coronal (curved arrows), sagittal (straight arrow), and lambdoid sutures (arrow heads); B, C: Of the cranium demonstrating bony bridging of bilateral squamosal sutures consistent with premature craniosynostosis. The approximate region of the normal squamosal sutures is indicated by straight arrows.



**Figure 2** Coronal computed tomography image of the brain demonstrating a local thumb printing/ beaten copper appearance (straight arrows) of the inner cortex in the region of bilateral squamosal fusion.

up for skull deformity.

## CASE REPORT

The patient was a 13 mo-old female who was born at 37 wk gestation and had a normal neonatal course. The patient's father carried a diagnosis of Crouzon syndrome. Her family history was otherwise unremarkable. Her past surgical history includes bilateral tear duct probing for bilateral nasolacrimal obstruction. On physical exam, the patient was noted to have macrocephaly (> 97% percentile for age) associated with a prominent forehead and relatively narrowed temples, proptosis, low set ears, orbital hypertelorism and the right cheek was higher and fuller than the left cheek. Given the family history of Crouzon syndrome and the patient's dysmorphic features, Crouzon syndrome was suspected. Genetic testing was done to confirm diagnosis and a mutation in the FGFR2 gene was noted. The patient was then referred to our radiology department to have a head CT performed to rule out craniosynostosis.

CT of the head without intravenous contrast was performed in our institution for further evaluation using the following technique: GE Medical Systems, Pitch: 0.625:1,

Pixel spacing: 0.400 mm/0.400 mm, Kvp 120, mA 109, rotate time: 0.8, ST 3.75 mm, FOV 20 cm, Tilt 0, W: 90, L: 40. Two dimensional images (2D) in the sagittal and coronal planes were reformatted at ST of 3 mm as well as three dimensional images of the skull (3D). The 3D images demonstrate normal appearance of the bilateral coronal and lambdoid sutures and normal appearance of the sagittal suture (Figure 1A). The metopic suture is not visualized, having normally fused. However, there is premature fusion of the squamosal sutures bilaterally (Figure 1B). Coronal 2D reformatted image (Figure 2) shows the local thumb printing (beaten copper) appearance of the inner cortex in the region of bilateral squamosal suture fusion.

The correction of the craniofacial deformities in a patient with Crouzon syndrome and bilateral squamosal craniosynostosis is accomplished through a series of operations over time with respect to the growth and development of the craniofacial skeleton. Around the first year of age, the patient will undergo subtotal cranial vault remodeling to release the fused sutures and correct the skull shape deformities. Subsequently, at around 7 years of age, the patient will be scheduled to undergo either a facial bipartition or Lefort III advancement to correct the hypertelorism or midface hypoplasia. At skeletal maturity, the patient will undergo orthognathic surgery to correct malocclusion when necessary.

## DISCUSSION

The development of the human calvaria is a complex process that is still not completely understood. There are 5 primary ossification centers which meet to form 6 main sutures. When this process occurs normally, it culminates to a normally shaped adult head<sup>[7]</sup>.

Craniosynostosis is a pathologic condition which is characterized by the premature fusion of cranial sutures which results in deformity of the vault and cranial base and is often associated with neurological sequelae secondary to alteration in cranial volume and restriction in brain growth. It may occur alone or in association with

other anomalies making up various syndromes<sup>[8]</sup>.

Craniosynostosis is estimated to affect approximately 1 in 2500 children<sup>[9,10]</sup>. Different etiologies of craniosynostosis include metabolic anomalies, teratogens, genetic anomalies, and abnormal intrauterine pressures<sup>[11]</sup>.

Among the craniosynostosis that are attributed to genetics, most have been associated with a mutation involving 1 of 3 fibroblast growth factor receptors (FGFRs) which include FGFR1, FGFR2, or FGFR3<sup>[11]</sup>. In the majority of cases, Crouzon syndrome is caused by mutations in the fibroblast growth factor 2 (FGFR2) gene on chromosome 10q26<sup>[1,12,13]</sup>. However, in patients with Crouzon syndrome and acanthosis nigricans, FGFR3 mutations have also been identified<sup>[14]</sup>. FGFR gene family mutations have also been found in patients with other syndromic craniosynostosis including Pfeiffer syndrome, Jackson-Weiss syndrome, Apert, and Saethre-Chotzen's syndromes<sup>[15,16]</sup>.

Our patient tested positive for FGFR2 mutation which is also found in Pfeiffer and Apert Syndromes<sup>[17]</sup>. Pfeiffer and Apert syndromes share common features with Crouzon syndrome including craniosynostosis, hypertelorism, proptosis, maxillary bone hypoplasia and autosomal dominance inheritance. However Pfeiffer and Apert syndromes are acrocephalosyndactyly syndromes associated with brachydactyly and syndactyly which our patient did not have and which helped in confirming the diagnosis of Crouzon syndrome<sup>[18,19]</sup>.

The gold standard imaging modality in diagnosing craniosynostosis is CT scan with 3D surface-rendered reconstruction<sup>[20]</sup>. MRI has no significant role in the diagnosis of craniosynostosis or facial anomalies associated with Crouzon syndrome as it has poor details to bones. MRI of the brain may be indicated in the future only if patient develops neurologic symptoms.

The sagittal suture is most commonly affected in non syndromic craniosynostosis and is estimated to be affected in 40%-60% of cases<sup>[21]</sup>. This is followed by the coronal suture that is affected in 20%-30% of the cases and the metopic suture in less than 10% of the cases<sup>[21]</sup>. The pattern of suture involvement determines the shape of the skull defect<sup>[21]</sup>. There have been more than 100 different types of syndromes which have been identified to be associated with craniosynostosis<sup>[9,10]</sup>.

Crouzon syndrome is one of the most common of the craniosynostosis syndromes<sup>[21]</sup>. It account for up to 4.8% of all cases of craniosynostosis<sup>[13]</sup>. The coronal and sagittal sutures are most commonly involved in Crouzon syndrome<sup>[13]</sup>.

There have only been a handful of cases of squamosal suture synostosis described in the surgery literature, the few described in Crouzon patients were always associated with other type of craniosynostosis<sup>[10,22]</sup>. This is the first reported case of isolated squamosal suture synostosis in a patient with Crouzon syndrome. The squamosal suture arches posteriorly from the pterion and connects the temporal squama with the inferior border of the parietal bone<sup>[10]</sup>.

The treatment of craniosynostosis syndromes requires a multidisciplinary team including plastic surgeons, pediatricians, ENT specialist, radiologists, neurosurgeons, and clinical genetic specialist<sup>[23,24]</sup>. Surgical correction is undertaken to correct craniofacial deformities and functional problems such as elevated intracranial pressure, airway obstruction, ocular exposure and malocclusion. Surgical techniques that are commonly utilized include fronto-orbital advancement or cranial vault remodeling in first year of life to correct craniosynostosis<sup>[25]</sup>. Hypertelorism is usually corrected with facial bipartition at age 7. Distraction osteogenesis is commonly used with midface osteotomies to correct midface hypoplasia as it has multiple advantages over conventional mid-facial advancement such as the monobloc or LeFort III. Distraction techniques have been shown to reduce operating time, reduce blood loss, reduce infection, eliminate need of bone graft, and allow for large advancement of the midface. Class III malocclusion which is commonly seen in syndromic craniosynostosis patients is corrected at skeletal maturity with a Lefort I advancement or double jaw surgery<sup>[26-32]</sup>.

In a conclusion, although squamosal suture craniosynostosis is an extremely rare entity, especially when associated with Crouzon syndrome, it is extremely important for the radiologist to be familiar with its radiological appearance in order to prevent misdiagnosis and delayed treatment.

## COMMENTS

### Case characteristics

Thirteen-month-old female patient with Crouzon syndrome was found to have isolated bilateral squamosal suture synostosis.

### Clinical diagnosis

The patient was noted to have macrocephaly associated with a prominent forehead and relatively narrowed temples, proptosis, low set ears, orbital hypertelorism and asymmetry in appearance of the cheeks.

### Differential diagnosis

Pfeiffer and Apert syndromes.

### Laboratory diagnosis

Mutation in the FGFR2 gene was noted.

### Imaging diagnosis

Computed tomography finding: The three dimensional (3D) images demonstrate normal appearance of the bilateral coronal and lambdoid sutures and normal appearance to the sagittal suture. However, there is bony bridging in the region of squamosal sutures bilaterally indicating premature synostosis. Coronal 2D reformatted image shows the local thumb printing (beaten copper) appearance of the inner cortex in the region of bilateral squamosal suture fusion.

### Treatment

The treatment of craniosynostosis syndromes requires a multidisciplinary team including plastic surgeons, pediatricians, ENT specialists, radiologists, neurosurgeons, and clinical genetic specialists. A series of corrective operations will be performed over time.

### Experiences and lessons

The authors report the unique case of isolated bilateral squamosal suture synostosis in a patient with Crouzon syndrome. Although bicoronal sutures are most commonly involved in patients with Crouzon syndrome, it is important to be familiar with this rare entity of craniosynostosis in order to prevent misdiagnosis or delayed surgical treatment.

### Peer review

This is a simple case report on a young patient with Crouzon syndrome. It is



concise, well written and well illustrated.

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