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Case report

Computed tomography findings of Crouzon syndrome: A case report [☆]

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ARTICLE INFO

Article history:

Received 5 December 2021

Revised 20 January 2022

Accepted 22 January 2022

Keywords:

Craniofacial dysostosis
Tomography X-ray computed
Child
Brain diseases
Crouzon syndrome
Craniosynostoses
Exophthalmos

ABSTRACT

Crouzon syndrome is a genetic condition characterized by a premature fusion of skull sutures resulting in head and facial deformities. Crouzon syndrome is usually suspected at birth through physical examination or in the antenatal period via ultrasonographic assessment. Once Crouzon syndrome is suspected, advanced imaging methods such as three-dimensional computed tomography must be requested, showing early signs of cranial sutures fusion. In this paper, we present a case of a six-year-old girl who was taken to a pediatrician control appointment due to abnormal facies. During the physical examination, a suspicion of Crouzon syndrome was raised. Therefore, a head computed tomography was requested, showing asymmetrical calvarium thickening, diffuse indentation of the inner table of the skull, and moderate hydrocephalus with a big cyst in the posterior fossa. Due to these findings, the patient was remitted to maxillofacial surgery for further evaluation; however, the medical appointment could not be achieved as a consequence of the poor medical insurance of the girl. This paper aims to describe and discuss the computed tomography findings of Crouzon syndrome.

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Introduction

Crouzon syndrome is a rare genetic condition with an autosomal dominant inheritance caused by a mutation in the fibroblast growth factor receptor-2 (FGFR-2) [1–3]. This mutation

[☆] Competing interests: The authors have no conflict of interests to disclose.

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leads to premature fusion of skull sutures, causing head and facial deformities [1–3]. Crouzon syndrome occurs in approximately 16.5 cases per million live births (1:60,000); therefore, it was initially considered the most common craniosynostosis syndrome [1,2,4]. However, the frequency of Muenke syndrome has been rising rapidly, taking the position as the most frequent craniosynostosis syndrome [1,2,4].

Crouzon syndrome is usually suspected at birth through physical examination or in the antenatal period via ultrasonographic assessment [2]. Once Crouzon syndrome is suspected, advanced imaging methods such as three-dimensional computed tomography (CT) must be requested, showing early signs of cranial sutures fusion [2]. Genetic tests must be ordered to perform the final diagnosis only in specific cases where the clinical and radiological findings are unclear [1].

The treatment of Crouzon syndrome requires a multidisciplinary approach accompanied by cranial decompression during the first year of life to reduce the elevated intracranial pressure [3]. An early diagnosis and treatment of the disease can improve the quality of life of these young patients and decrease complications such as intracranial hypertension. This paper aims to discuss the CCTTCT findings of a patient with Crouzon syndrome in which poor medical insurance interfered with a prompt diagnosis and treatment.

Case description

A six-year-old girl was taken to the pediatrician due to complaints regarding abnormal facies. The patient had had a history of severe pectus excavatum, sleep apnea, congenital subglottic stenosis, and Dandy-Walker syndrome with recurrent hospitalizations since birth due to multiple episodes of seizures and non-communicating hydrocephalus that required the placement of a ventriculoperitoneal valve. On physical examination, an aware patient with mental impairment, short size (1 meter), low weight (12.7 kg), hypertelorism, proptosis, beaked nose, micrognathia, and retrognathia with no signs of dental, hearing, or digital malformations was evident (Fig. 1). The mother gave birth at 37 weeks through an elective cesarean section with no complications during pregnancy. No similar cases were described in the family.

During the physical examination, a suspicion of Crouzon syndrome was raised. Therefore, a head CT was requested, showing asymmetrical calvarium thickening, diffuse indentation of the inner table of the skull, and moderate hydrocephalus with a big cyst in the posterior fossa (Fig. 2). A three-dimensional CT reconstruction of the head was performed, showing an irregular skull, with a total fusion of both coronal sutures and partial fusion of the posterior region of the sagittal suture (Fig. 3). These findings correlate with Crouzon syndrome, chronic intracranial hypertension, and Dandy-Walker syndrome with moderate hydrocephalus.

Due to the physical examination and radiographic findings, Crouzon syndrome was diagnosed; therefore, the patient was

referred to maxillofacial surgery for further evaluation. However, the medical appointment could not be achieved as a consequence of the poor medical insurance of the girl.

The patient's prognosis was good six months after the diagnosis of Crouzon syndrome. No limitations derived from the craniofacial deformity were presented. The patient continued with language disabilities due to the brain affections caused by the Dandy-Walker syndrome.

Discussion

Syndromic craniosynostosis are caused by mesenchyme and ectoderm malformations. The ectoderm covers a vital role in brain embryogenesis; therefore, its alteration in craniosynostosis syndromes can lead to brain anomalies such as schizencephaly or Dandy-Walker syndrome, as shown in this case [5–9]. Several mutations in transcription-derived growth factors have been associated with craniosynostosis syndromes, such as TWIST, FBN1, FGFR 1, 2, 3, tumor growth factor-b1, and b2 [10]. Particularly, Crouzon syndrome is caused by mutations in FGFR 2 or FGFR 3 [10].

FGFR 2 and FGFR 3 mutation cause most Crouzon syndrome manifestations, such as craniosynostosis, hypertelorism, and intracranial volume reduction [9,11,12]. Specifically, FGFR 3 mutation is associated with acanthosis nigricans and higher incidence of hydrocephalus [1]. This patient presented with hydrocephalus but did not show acanthosis nigricans; therefore, it is unlikely that the patient had an FGFR 3 mutation, but instead, it is expected an FGFR 2 alteration. In addition, based on previous literature, FGFR 2 mutation is considered the most common type of genetic alteration in patients with Crouzon syndrome [11].

Due to a lack of resources in our context, we did not carry out any genetic test to diagnose Crouzon syndrome; instead, we performed the diagnosis based on clinical and radiological findings [1]. It has been described that genetic test for Crouzon syndrome are only required if the clinical or radiological presentation is unclear [1].

Craniosynostosis are characterized radiologically by the fusion of the skull sutures, usually compromising either the coronal (20%-25%), metopic (5%-15%), lambdoid (<5%), and sagittal (40%-55%) sutures [11–14]. This explains the typical midface hypoplasia and the upper airway obstruction presented in these individuals [11–14]. Our patient had a tendency toward the early closure of the coronal and sagittal sutures.

Head CT is helpful in patients with Crouzon syndrome to assess the status of skull sutures, offering several benefits over X-ray and ultrasound. The main advantage of head CT is offering a more accurate representation of the patient's skull anatomy using three-dimensional reconstructions and the capacity to assess the brain morphology [1,9,10,12,15]. Magnetic resonance imaging is not frequently used to evaluate the skull in Crouzon syndrome; instead, it is the preferred method to



Fig. 1 – Physical examination showing hypertelorism (red box), proptosis (red arrow), beaked nose (blue arrow), micrognathia, and retrognathia (purple arrow).

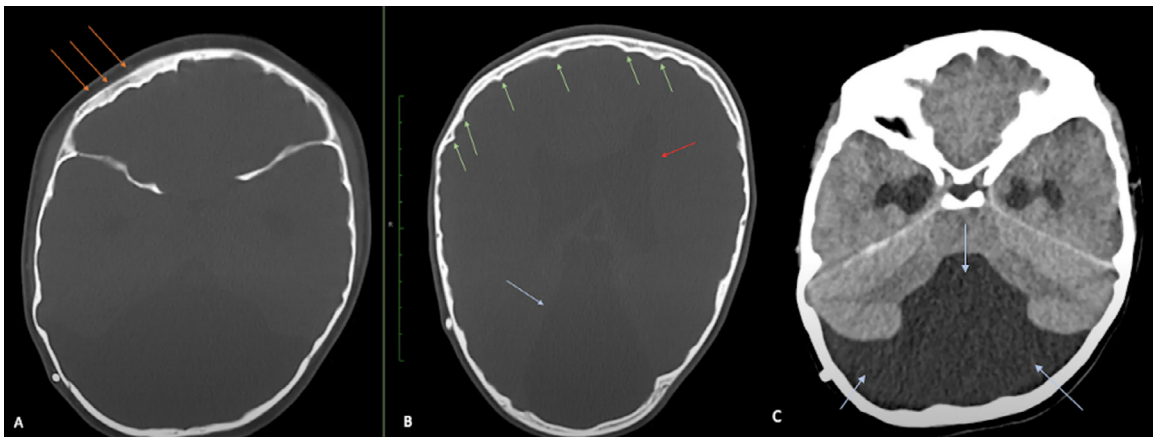


Fig. 2 – Non-contrast axial head CT in bone window shows asymmetrical calvarium thickening in the right frontal bone (orange arrows) (A). Moderate hydrocephalus is seen (red arrow) (B). A big cyst located in the posterior fossa (blue arrows), and diffuse indentation of the inner table of the skull are evident (green arrows) (B, C).

assess the brain parenchyma when CT images show incidental findings [10,12].

In this case, several factors could have contributed to the genesis of intracranial hypertension. The first is the fusion of the coronal and sagittal sutures, which could have restricted the expansion of the skull while the brain was continually growing. The second is the presence of a big posterior fossa cyst, which could have contributed to intracranial hypertension due to mass effect within an already restricted skull vault. This rise in the intracranial pressure led to moderate hydrocephalus and diffuse indentation of the inner table of the skull documented by CT. The indentation of the internal table of the skull represents the well-known “copper beaten skull sign” but detected in CT [16].

It is essential to mention that fused sutures tend to have an increased bone deposition, leading to para-sutural sclero-

sis and bony ridges [13]. Our patient showed an asymmetrical calvarium thickening, suggesting bony ridges formation adjacent to the coronal suture.

All craniosynostosis syndromes can produce similar radiological and clinical characteristics. Therefore, identifying Crouzon syndrome, among others syndromic craniosynostosis, can result in a diagnostic challenge. The lack of hands and foot anomalies is crucial to distinguish Crouzon syndrome from other craniosynostoses such as Apert syndrome, Pfeiffer syndrome, Saethre-Chotzen syndrome, and Muenke syndrome [16]. This patient did not have hands or feet abnormalities, which aligns with the clinical presentation of Crouzon syndrome described in the literature [16].

The treatment of Crouzon syndrome is performed by a multidisciplinary group based on the severity, functionality, and appearance-related needs of the patient [17,18]. The treat-

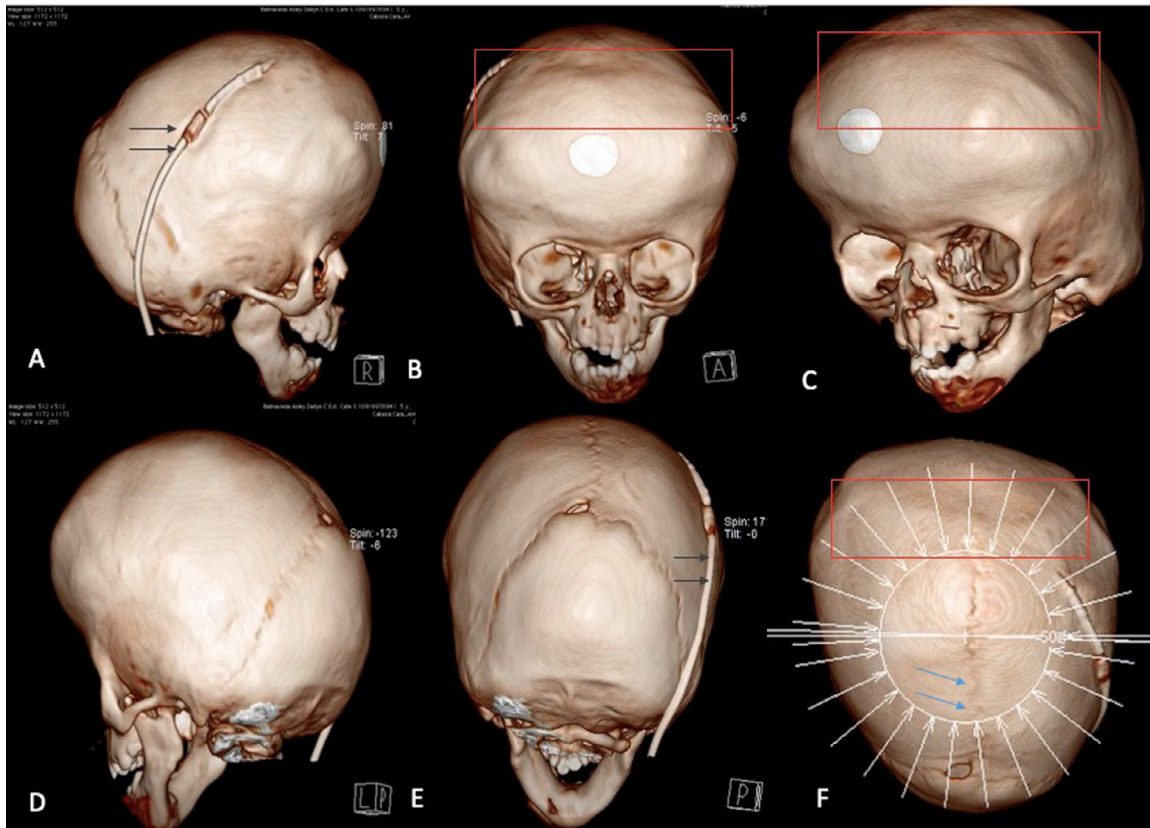


Fig. 3 – Three-dimensional CT skull reconstruction. An increased diameter of the head is evident (A–F). A correctly placed ventriculoperitoneal valve is shown (black arrow) (A, E). The total fusion of both coronal sutures is appreciated (red box) (B, C, F). Partial fusion of the posterior region of the sagittal suture is seen (blue arrow) (F). Lambdoid sutures are normal.

ment consists of frontal-orbital advancement and cranial decompression to prevent the rise of intracranial pressure and other complications related to the syndrome [17,18]. Ideally, the treatment should begin during the first year of life; therefore, it is crucial to perform a prompt diagnosis. In this case, Crouzon syndrome diagnosis was performed late; therefore, the only therapy established was the management of the hydrocephalus and its complications, but no treatment was settled for her facial deformities.

Conclusions

Crouzon syndrome can be diagnosed based on clinical and radiological findings; therefore, genetic tests are only required when the diagnosis is unclear. Head CT is the primary radiological method to assess patients with Crouzon syndrome by showing alterations in the skull such as fusion of the sutures, asymmetrical calvarium thickening, diffuse indentation of the inner table of the skull, in addition to allowing a complete evaluation of the brain. Physicians need to be aware of the possible etiological association between Crouzon syndrome and Dandy-Walker syndrome.

Patient consent

Patient consent has been obtained.

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