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Case Report

Crouzon Syndrome with unilateral blindness: A Case from Benguela, Angola

Síndrome de Crouzon y ceguera unilateral: reporte de un caso en Benguela, Angola

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Abstract

Background: Crouzon syndrome is a rare genetic craniosynostosis caused by mutations in the fibroblast growth factor receptor 2 gene (*FGFR2*), leading to premature suture fusion and characteristic craniofacial deformities. It is usually inherited in an autosomal dominant pattern with variable expression. **Case Presentation:** We describe a 15-year-old male from Benguela, Angola, presenting with progressive bilateral proptosis, cranial deformities, and unilateral visual loss. Computed tomography confirmed premature fusion of the coronal and sagittal sutures and shallow orbits, consistent with Crouzon syndrome. Ophthalmologic examination showed corneal leukoma and optic neuropathy in the right eye. Orbital decompression was indicated; however, irreversible blindness had already occurred due to delayed diagnosis. **Conclusion:** Early detection and coordinated multidisciplinary intervention involving ophthalmology, craniofacial surgery, genetics, and psychology are vital to improving outcomes. Reporting such cases from Africa enhances clinical awareness and underscores the need to strengthen diagnostic and surgical capacities for craniofacial disorders in developing regions.

Keywords: Crouzon Syndrome; Craniofacial Dysostosis; Exophthalmos; Angola (Source: MeSH, NLM).

Resumen

Antecedentes: El síndrome de Crouzon es una craneosinostosis genética rara causada por mutaciones en el gen del receptor del factor de crecimiento de fibroblastos 2 (FGFR2), que conduce a la fusión prematura de las suturas craneales y a deformidades craneofaciales características. Generalmente se hereda con un patrón autosómico dominante y presenta expresión variable. **Presentación del caso:** Se describe el caso de un varón de 15 años procedente de Benguela, Angola, que presentó proptosis bilateral progresiva, deformidades craneales y pérdida visual unilateral. La tomografía computarizada confirmó la fusión prematura de las suturas coronal y sagital, así como órbitas poco profundas, hallazgos compatibles con síndrome de Crouzon. El examen oftalmológico evidenció leucoma corneal y neuropatía óptica en el ojo derecho. Se indicó descompresión orbitaria; sin embargo, la ceguera irreversible ya se había establecido como consecuencia del diagnóstico tardío. **Conclusión:** La detección precoz y la intervención multidisciplinaria coordinada, que involucre oftalmología, cirugía craneofacial, genética y psicología, son fundamentales para mejorar los resultados clínicos. El reporte de este tipo de casos en África contribuye a aumentar la conciencia clínica y resalta la necesidad de fortalecer las capacidades diagnósticas y quirúrgicas para los trastornos craneofaciales en regiones en desarrollo.

Palabras clave: Disostosis Craneofacil; Exoftalmia; Angola (Fuente: DeCS, BIREME).

Introduction

Crouzon syndrome is one of the most well-known and extensively studied forms of syndromic craniosynostosis. It represents a rare genetic condition characterized by premature fusion of cranial sutures, leading to typical craniofacial deformities, and is frequently associated with mutations in the fibroblast growth factor receptor 2 (FGFR2) gene [1] [2]. First described by Octave Crouzon in 1912, the syndrome follows an autosomal dominant inheritance pattern with variable penetrance and heterogeneous clinical expression [3]. The global incidence is estimated to range between 1 in 25,000 and 1 in 60,000 live births, with no predilection for sex or ethnic group [4].

The most characteristic clinical manifestations include exophthalmos, maxillary hypoplasia, ocular proptosis, strabismus, beak-shaped nose, and an apparent increase in cranial vault height [5]. In some cases, hearing impairment, sleep apnea, and respiratory compromise due to midfacial hypoplasia may occur [6]. Despite the evident aesthetic impact, cognitive development is usually preserved, distinguishing it from other syndromic craniosynostoses such as Apert and Pfeiffer syndromes [7].

In the Benguela region, in southern Angola, access to specialized pediatric neurosurgical and craniofacial surgical care remains limited. Consequently, the documentation and dissemination of clinical case reports are particularly valuable for the African medical literature. Case reports originating from African contexts make important contributions to local epidemiological understanding, stimulate the development of multidisciplinary teams, and support the creation of referral and counter-referral protocols adapted to regional conditions [8].

This case report aims to describe an uncommon presentation of Crouzon syndrome in an Angolan adolescent, highlighting the diagnostic process, distinctive ocular manifestations, and multidisciplinary management approach in a resource-limited setting.

Case presentation

This case concerns a 15-year-old male adolescent, born and residing in Benguela, Angola, who presented to the Benguela Ophthalmologic International Center (Centro Oftalmológico Internacional de Benguela, COIB) on August 5, 2025. The main reason for consultation was a progressive bilateral increase in ocular volume, associated with ocular pain, frontal headache, and family concern due to the evident craniofacial deformity **(Figure 1)**.

The patient was born to a 35-year-old mother after a full-term pregnancy with an uncomplicated hospital vaginal delivery. The mother attended three prenatal consultations, during which obstetric ultrasounds showed no structural abnormalities. There were no reported complications during pregnancy or delivery. Immunizations were up to date, and the parents denied a family history of similar conditions, although the autosomal dominant nature of the disorder raises the possibility of a de novo mutation.

According to the parents, since the first year of life, the patient exhibited a gradual increase in eyeball prominence associated with persistent nasal discharge, which led to his

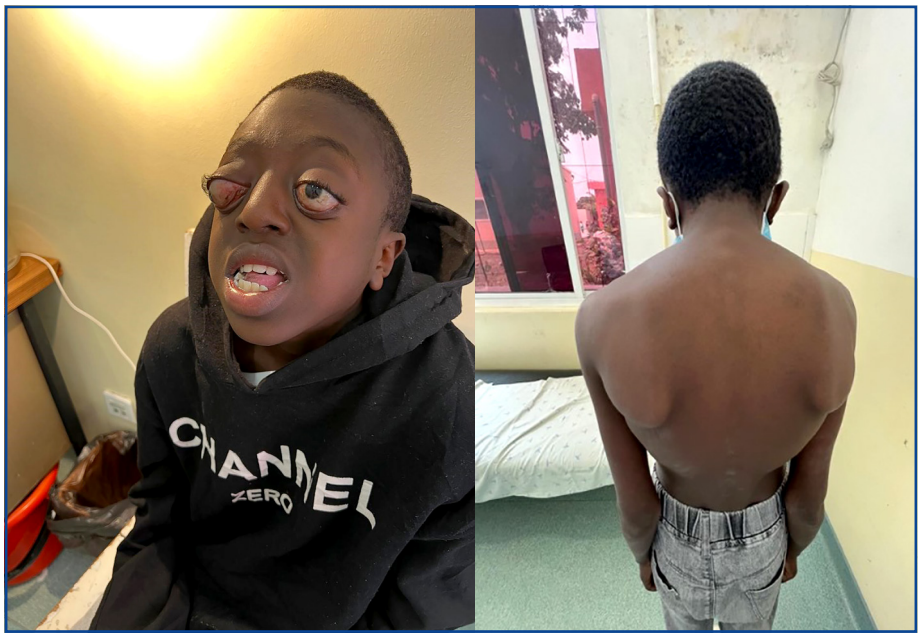

Figure 1. Patient's face showing characteristic bilateral ocular proptosis and craniofacial deformity.

Source: Authors' own clinical documentation, 2025, with informed consent.

first medical evaluation at the COIB. At that time, a congenital craniofacial syndrome was hypothesized, and annual follow-up was advised to monitor craniofacial development.

During childhood, the patient experienced multiple hospital admissions due to episodes of respiratory distress attributed to abnormal midfacial anatomy. At the age of four, the family traveled to the Republic of Namibia seeking specialized assessment, where symptomatic medications unspecified by the relatives were prescribed.

The clinical condition remained stable throughout early childhood. However, during adolescence, there was a marked progression of ocular protrusion and the onset of visual disturbances, prompting a new referral to the COIB for further evaluation and management.

General Physical Examination

The patient presented with poor nutritional status and mild restriction of cervical movements. Cranial deformities compatible with premature suture fusion were observed, including turricephaly and scaphocephaly. Marked bilateral proptosis, widened palpebral fissures, fish-like mouth, auricular asymmetry, short neck, and evident dental malocclusion were noted **(Figure 2)**. Vital signs were stable: blood pressure 120/70 mmHg, heart rate 80 bpm, respiratory rate 24 cpm, and temperature 36.5°C.

Ophthalmologic Examination

Ophthalmic assessment revealed increased eyelid elasticity, reduced ocular motility, and decreased visual acuity in the right eye (counting fingers at 1 meter), while functional vision was preserved in the left eye. Examination of the ocular media showed corneal opacity due to exposure in the right eye and preserved transparency in the left. The anterior segment of the right eye exhibited corneal leukoma with neovascularization, whereas no structural alterations were observed in the left eye.

Fundoscopic evaluation demonstrated attached retinae, optic nerve heads with temporal pallor, preserved physiological cupping, and maculae with intact foveal reflexes **(Figure 3)**. These findings were consistent with optic neuropathy secondary to cranio-orbital compression, a characteristic feature of the syndrome.

Complementary Examinations

Laboratory tests showed hemoglobin of 11.3 g/dL with no significant leukocytic abnormalities. Serologic tests for HIV, hepatitis B, and hepatitis C were negative. Craniofacial computed tomography revealed premature fusion of the coronal and sagittal sutures, shallow orbits, and marked hypertelorism, findings typical of Crouzon syndrome (**Figure 4**). Optical coherence tomography (OCT) and fundus photography confirmed anterior segment changes compatible with chronic corneal exposure **(Figure 5)**.

Diagnostic Discussion

The constellation of clinical and radiological findings supported the diagnosis of Crouzon syndrome, also known as craniofacial dysostosis, a disorder of autosomal dominant inheritance associated with mutations in the *FGFR2* gene located on chromosome 10. Differential diagnoses included

Figure 2. Lateral view demonstrating turricephaly and “fish-mouth” appearance (right) and a short neck with auricular asymmetry (left).

Source: Authors’ own clinical documentation, 2025, with informed consent.

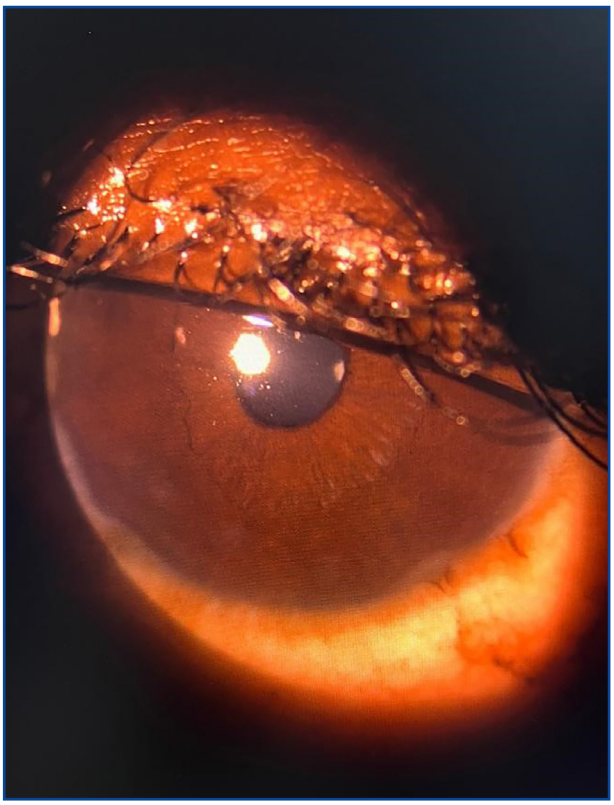

Figure 3. Anterior segment of the right eye showing corneal leukoma and neovascularization.

Source: Authors' own clinical documentation, 2025, with informed consent.

Apert and Pfeiffer syndromes, which share overlapping phenotypic traits but differ in their genetic and skeletal characteristics.

Management and Follow-up

The case was referred to a multidisciplinary team comprising ophthalmology, craniofacial surgery, medical genetics, otorhinolaryngology, and clinical psychology. Due to the severity of the proptosis and the corneal opacity, orbital decompression surgery was proposed to reduce intraorbital pressure and prevent ocular extrusion. Despite therapeutic interventions, irreversible blindness in the right eye was established, resulting from prolonged exposure keratopathy and compressive optic neuropathy. Continuous follow-up and psychosocial support were deemed essential to minimize the functional and emotional repercussions of the disease.

Discussion

Crouzon syndrome, first described in 1912 by the French neurologist Octave Crouzon [9], is a rare genetic disorder resulting from mutations primarily affecting the FGFR2 gene, located on chromosome 10q25–10q26, leading to abnormal cranial suture development and premature ossification [2][3][10]. Clinically, the condition is characterized by early fusion of two or more cranial sutures, most commonly the coronal and sagittal

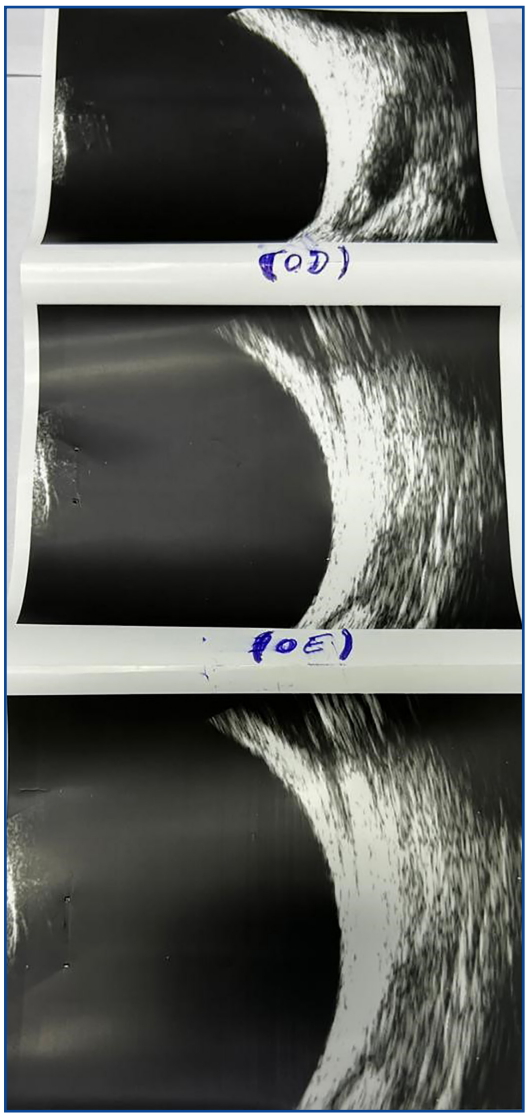

Figure 4. Cranial computed tomography showing fusion of the coronal and sagittal sutures and shallow orbits.

Source: Authors' own clinical documentation, 2025, with informed consent.

sutures, resulting in restricted cranial growth and compensatory craniofacial deformities [6][9].

In Angola, Crouzon syndrome was first reported by Quilezi *et al*. in 2021, describing a female infant diagnosed at Benguela General Hospital who presented with typical craniofacial manifestations, including exophthalmos, craniosynostosis, hypertelorism, and cleft palate [11]. This highlights the rarity of documented cases in the region and reinforces the importance of reporting cases from resource-limited settings to expand epidemiological knowledge and clinical awareness [8].

The typical clinical manifestations of Crouzon syndrome include acrocephaly, midface hypoplasia, "parrot-beak" nose, shallow orbits, proptosis, strabismus, dental malocclusion, and, in advanced cases, optic nerve compromise [1][6][9]. Computed tomography usually demonstrates premature

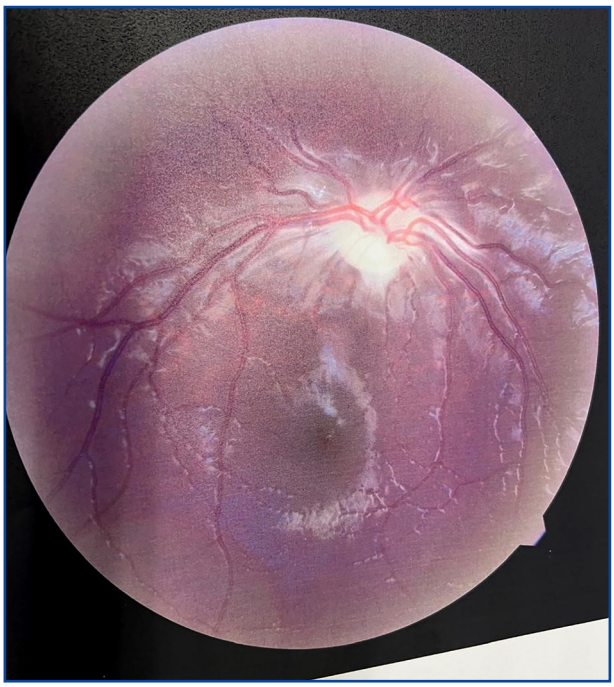

Figure 5. Retinography (advanced ocular complication, right eye blindness, and risk of globe luxation/expulsion).

Source: Authors' own clinical documentation, 2025, with informed consent.

fusion of cranial sutures and abnormal cranial vault morphology, explaining the anterior displacement of the orbits and the increased risk of ocular exposure and visual impairment [6][9]. In the present case, the marked bilateral proptosis was associated with severe ocular complications, culminating in irreversible unilateral blindness, most likely due to prolonged corneal exposure and compressive optic neuropathy, which are well-recognized complications in untreated or late-treated craniosynostosis [1][6][9].

From a genetic perspective, mutations in FGFR2 promote constitutive activation of fibroblast growth factor signaling, stimulating premature osteoblastic differentiation and early suture fusion [3][10]. Although molecular confirmation was not possible in this case due to limited local resources, the clinical phenotype strongly supports a pathogenic FGFR-related mutation. The absence of a family history is consistent with the high proportion of sporadic, de novo cases described in the literature [2][7]. These findings underscore the relevance of genetic counseling, even in families without previous affected members, although access to genetic services remains limited in many low-income countries [2][10].

Surgical management remains the cornerstone of treatment and aims to correct cranial deformities, relieve intracranial pressure, and preserve visual and neurological function [5][6][9]. The surgical strategy is usually staged, beginning with cranial vault remodeling in early childhood, followed by midface and orbital corrections when necessary [5][9]. In this case, orbital decompression was indicated to reduce intraorbital pressure and prevent further optic damage; however, delayed diagnosis significantly limited the therapeutic benefit. Long-term follow-up requires periodic neurological, ophthalmological, and craniofacial assessments, often supported by imaging studies to monitor cranial growth and potential complications [6][9].

Multidisciplinary management is essential due to the complex functional and psychosocial implications of the syndrome [6][9]. The involvement of craniofacial surgeons, ophthalmologists, pediatricians, dentists, and speech therapists is fundamental to optimize outcomes and quality of life. Facial deformities may significantly impact self-esteem and social integration, particularly during adolescence, reinforcing the importance of psychological support as part of comprehensive care [8][9].

In developing countries such as Angola, delayed diagnosis is frequently related to limited access to specialized imaging, scarcity of trained professionals, and restricted availability of genetic testing. These constraints contribute to worse functional outcomes, particularly visual impairment and irreversible sequelae. Nevertheless, this case demonstrates that careful clinical evaluation remains a powerful diagnostic tool, even in resource-limited settings. Early recognition, timely referral, and integrated management are critical to minimizing complications and improving long-term prognosis. Furthermore, the documentation of cases from African contexts contributes to strengthening scientific evidence and guiding health policies adapted to regional realities [8].

Conclusion

This case highlights the clinical, ophthalmologic, and radiologic features of Crouzon syndrome in an adolescent patient diagnosed at a late stage in Benguela, Angola. The clinical presentation, characterized by severe bilateral proptosis, cranial deformities, and irreversible unilateral blindness, illustrates the consequences of delayed diagnosis and limited access to specialized multidisciplinary care. Despite the absence of molecular confirmation, the phenotype was consistent with FGFR2-related craniosynostosis, reinforcing the diagnostic value of detailed clinical and imaging evaluation in resource-limited settings.

The case underscores the importance of early recognition and multidisciplinary management, encompassing ophthalmologic, craniofacial, genetic, and psychological care to prevent functional impairment and improve patients' quality of life. Furthermore, it emphasizes the need to strengthen diagnostic and surgical capacities in low-income countries and to promote the inclusion of craniofacial anomalies in neonatal and pediatric screening programs. Reporting such cases contributes valuable insights to the regional and global literature, fostering awareness, improving clinical vigilance, and guiding the development of locally adapted health strategies for syndromic craniosynostoses.

Additional Information

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Data availability: The data supporting the findings of this case report are available within the article. Additional details may be provided upon reasonable request to the corresponding author.

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