

## Crouzon syndrome

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### KEYWORDS:

Crouzon syndrome;  
Papilledema;  
Proptosis;  
Craniofacial  
syndrome;  
Craniosynostosis

### Abstract

**BACKGROUND:** Crouzon syndrome is a rare genetic disorder characterized by distinctive malformations of the skull and facial region. Premature cranial suture closure is the most common skull abnormality. Optic disc edema and proptosis are among the most common ocular findings.

**CASE REPORT:** We present a case of a 5-year-old girl with Crouzon syndrome displaying classic facial abnormalities along with proptosis and papilledema. The child's condition was improved dramatically after a monoblock advancement procedure.

**CONCLUSIONS:** The differential diagnosis of the condition and treatment options are discussed. The referring optometrist can play an integral role in the multidisciplinary care the patients require.  
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Crouzon syndrome is a rare genetic disorder that may be evident at birth or during infancy. The disorder is characterized by distinctive malformations of the skull and facial (craniofacial) region. Such abnormalities may vary greatly in range and severity from case to case, including variations among affected family members. However, in most infants with Crouzon syndrome, the fibrous joints between the cranial sutures close prematurely (craniosynostosis). In addition, facial abnormalities typically include proptosis owing to shallow orbits; divergent strabismus or exotropia; ocular hypertelorism; and a small, underdeveloped upper jaw (hypoplastic maxilla), with protrusion of the lower jaw (relative mandibular prognathism). Multiple staged surgeries are the general treatment plan for patients with Crouzon syndrome. With proper treatment, these patients can be productive and active members of mainstream society.

### Case report

A 5-year-old girl presented to the office complaining of ocular redness and irritation of a long-standing duration in both eyes (OU). The child's mother stated that the girl "constantly rubs" her eyes and complains of burning. Review of systems was unremarkable; specifically, the mother reported normal labor and delivery as well as normal developmental milestones. There were no anomalies in any siblings or near relatives reported. The child was not on any medications and denied any medical allergies. Her last ocular examination was about 3 years prior, at which time the mother reported, "everything was normal."

Best-corrected visual acuities were 20/30 in the right eye (O.D.) and 20/40 in the left eye (O.S.). External examination found gross proptosis that measured 20 mm on exophthalmometry. The child displayed a flattened bridge and dental malocclusion. The child had incomplete lid closure. Interpupillary distance was 68 mm. Her older brother was present, and had a normal facial appearance, as did the mother and father. Versions were full and smooth without any underactions or overactions of the intraocular muscles. Visual fields were full to finger count confrontation. The child showed gross stereopsis with randot animals. Color

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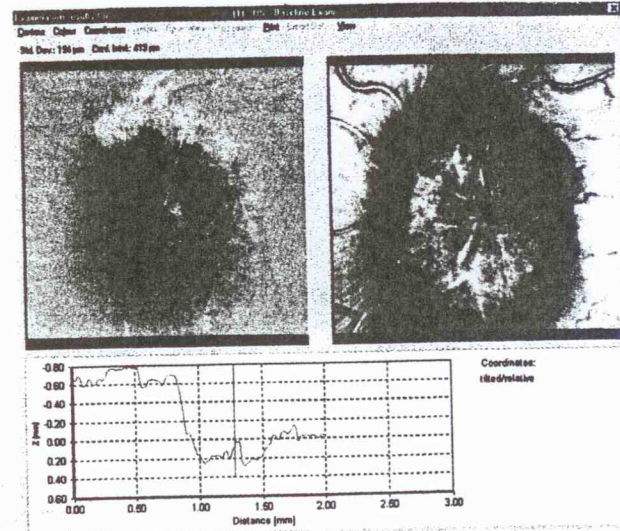
**Figure 1** Patient at initial presentation. Note prominent globes.

vision testing was normal in either eye by pseudoisochromatic plates. Pupils were equally round and reactive to light and accommodation without afferent pupillary defect. Slit lamp examination found a moderate interpalpebral conjunctival injection with some small ulcerations on both the nasal and temporal aspects of the bulbar conjunctiva and 360 degrees of diffuse superficial punctate keratitis OU. Intraocular pressures were 15 mmHg OU by Goldmann applanation. Dilated fundus examination found bilateral optic disc edema. Threshold visual fields were attempted, which the child was unable to perform. She was able to complete a 76-point screening field, which displayed some peripheral changes OU (see Figure 1).

The findings were discussed with the child's parents. The child's physical appearance was so strikingly different from other family members, and with the presence of exposure keratopathy and bilateral disc edema, the question was raised if anyone had ever discussed the diagnosis of a craniofacial syndrome.

Treatment of her presenting conditions consisted of non-preserved artificial tears hourly and polytrim ophthalmic ointment. The child was referred immediately to an ophthalmologic consultation center for evaluation of the optic disc edema, where Heidelberg retinal tomography (HRT) was attempted OU, but only O.S. obtained (see Figure 2).

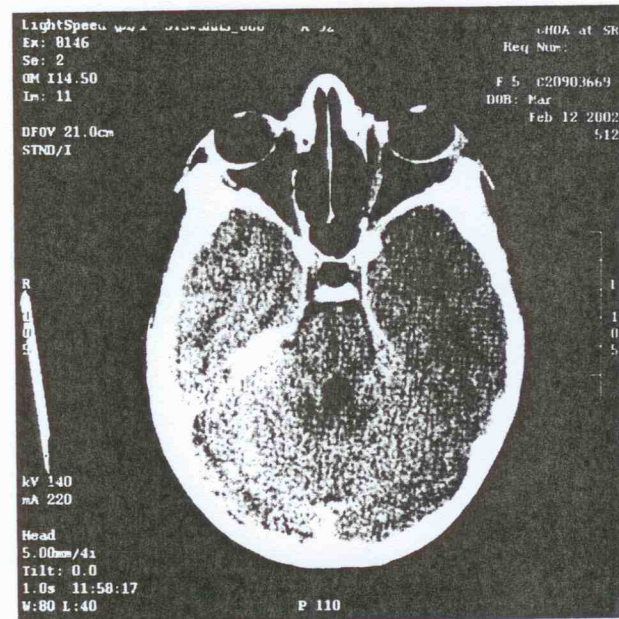
In addition, a referral was made to the Children's Hospital of Atlanta, Center for Craniofacial Disorders for evaluation. There she underwent a battery of tests, including audiology (which was within normal limits), sleep study to evaluate obstructive apnea (a positive test), and genetics evaluation. Medical evaluation found a dysmorphic facies with maxillary hypoplasia, proptosis, and evidence of bicoronal synostosis, all clinical findings consistent with



**Figure 2** HRT II analysis of patient's left eye. Note optic disc edema.

Crouzon syndrome. Family evaluation found that because "neither parent has any clinical features of (Crouzon), the greatest likelihood is that this represents a de novo mutation." Head computed tomography (CT) scan with 3-dimensional (3D) reconstruction showed bicoronal synostosis and evidence of chronically elevated intracranial pressure. Figure 3 shows the CT scan of the orbits displaying marked proptosis of both globes. Figure 4 shows CT scan of skull displaying premature suture closure.

She underwent combined craniofacial and neurosurgical intervention 2 months after her initial presentation. A monoblock advancement procedure, which opened the closed sutures while advancing the hypoplastic maxilla and orbit was performed using 1-stage resorbable bone distractor. Intraoperative cerebrospinal fluid (CSF) pressure measure-



**Figure 3** CT scan of orbits show marked proptosis OU.

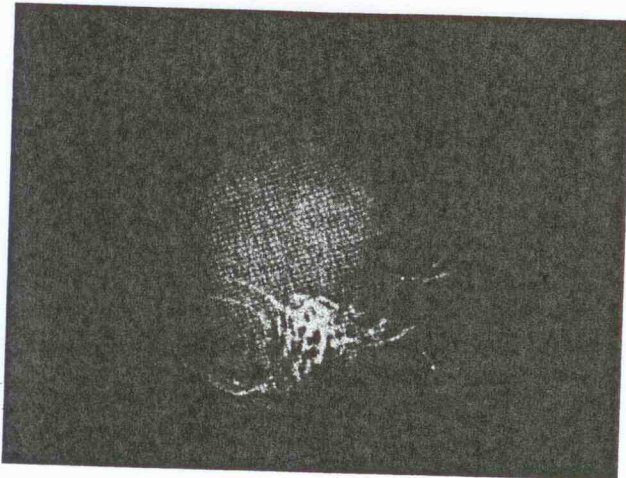


Figure 4 CT scan of skull. Note absence of suture lines.

ments confirmed the radiographic impression of increased intracranial pressure. A total of 20 mm of advancement was achieved, relieving the intracranial pressure and restoring normal facial and orbital morphology as well as occlusion. She tolerated the procedure well and is being followed up by the neurosurgeon at regular intervals. She was returned to the office for followup of the papilledema, which was present but minimal at the 4-month postoperative visit. Visual field defect was no longer present on screening fields. Her uncorrected visual acuity at last visit was 20/20 OU. Figure 5 shows the patient following monoblock expansion advancement surgery.

## Discussion

### Background

In 1912, Crouzon described the hereditary syndrome of craniofacial dystosis in a mother and son.<sup>1</sup> He described the triad as skull deformities, facial anomalies, and exophthalmos. Premature craniosynostosis, midfacial hypoplasia, and exophthalmos form the triad now known as Crouzon syndrome.<sup>2</sup> Crouzon syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity.<sup>3</sup> It is characterized by premature closure of cranial sutures, most commonly the coronal and sagittal sutures, resulting in abnormal skull growth and affecting growth and development of the orbits and maxillary complex. Other clinical features include hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, hypoplastic maxilla, and relative mandibular prognathism.<sup>4</sup> Unlike some other forms of autosomal dominant craniosynostosis, there are no digital abnormalities.<sup>5</sup> Acanthosis nigricans (a disorder causing velvety, light-brown-to-black markings usually on the neck, under the arms, or in the groin) is the main dermatologic manifestation of Crouzon syndrome.<sup>6</sup>

## Characteristics

Crouzon syndrome patients have 3 distinct features<sup>4</sup>:

- Craniosynostosis, most often of the coronal, and occasionally lambdoid or sagittal sutures
- Underdeveloped midface with receded cheekbones or exophthalmos
- Ocular proptosis, which is caused by very shallow orbits. The patient may have strabismus and hypertelorism.

Some other features commonly seen in these patients are visual disturbances related to an imbalance of the extraocular muscles and hearing loss owing to recurrent ear infections. The mental capacity of Crouzon syndrome patients is usually in the normal range; however, some mental delay has been reported that may be related to increased intracranial pressure.<sup>7</sup>

## Pathophysiology

Crouzon syndrome is caused by mutations in the fibroblast growth factor receptor-2 (*FGFR2*) gene, which is mapped to chromosome locus 10q25-10q26.<sup>8</sup> Fifty percent of incidents of Crouzon syndrome are not inherited and are the result of new mutations.

Premature synostosis of the coronal and sagittal and, in some cases, lambdoid sutures begins in utero and is manifest at birth. The order and rate of suture fusion determine the degree of deformity and disability. Once a suture becomes fused, growth perpendicular to that suture becomes restricted, and the fused bones act as a single bony structure. Compensatory growth occurs at the remaining open sutures to allow continued brain growth, resulting in abnormal bone growth and producing facial deformities. When multiple sutural synostoses occurs, it is likely to initiate to premature



Figure 5 Patient after monoblock expansion advancement surgery.

