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Crouzon syndrome

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KEYWORDS:
Crouzon syndrome;
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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.

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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 nonpreserved 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 Heidelburg 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

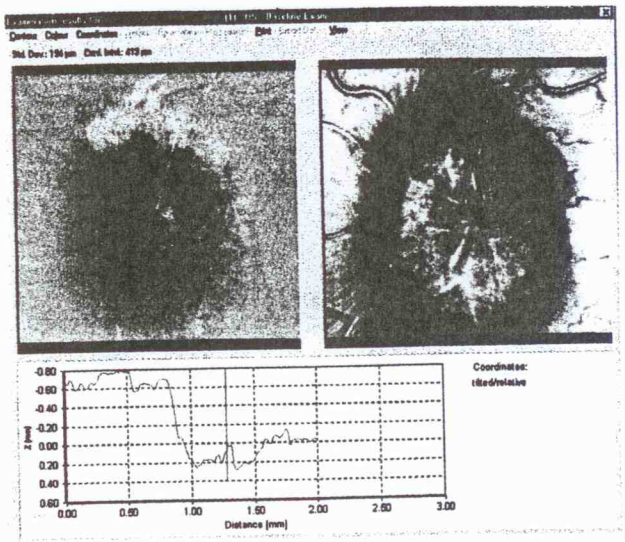

Figure 2 HRT II analysis of patient's left eye. Note optic disc edema

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 monobloc advancement procedure, which opened the closed sutures while advancing the hypoplastic maxilla and orbits was performed using 1-stage resorbable bone distractors. Intraoperative cerebrospinal fluid (CSF) pressure measure

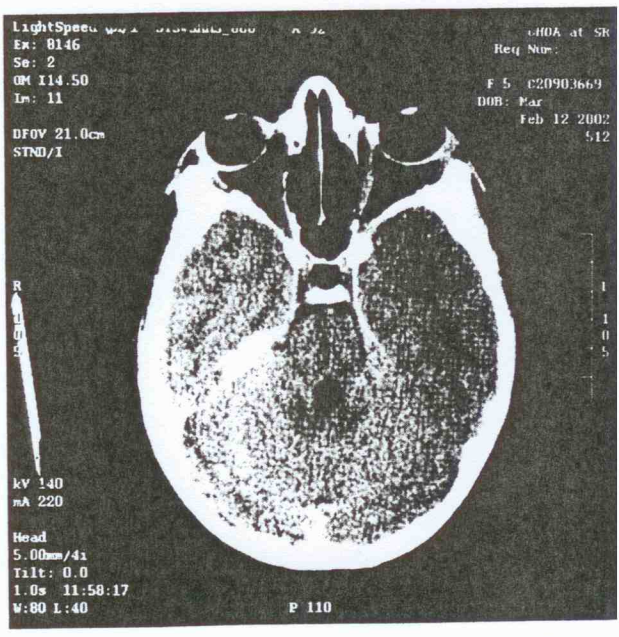

Figure 3 CT scan of orbits show marked proptosis OU.