Crouzon Syndrome: Visual Diagnosis

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CASE PRESENTATION

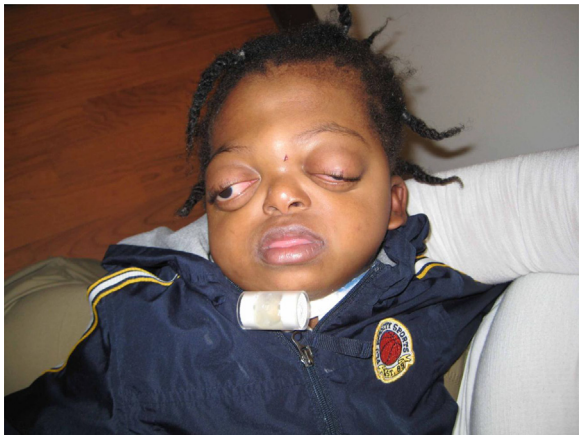
A 5-month-old Black male infant presented as a new patient to our primary care pediatric clinic. His maternal grandmother reported that he had been in her care since 1 month of age. She was concerned about his eyes, which had appeared to be bulging since birth. He was unable to close his eyes while sleeping (Figure 1), and she had been instilling lubricating ointment at bedtime to prevent keratoconjunctivitis sicca (eye dryness). He also had an abnormally shaped head, a café-au-lait spot, and a hemangioma on the medial aspect of the right anterior lower leg.

The patient was born after a 32-week gestation complicated by maternal tobacco, alcohol, and illicit drug use. The mother had prenatal care only in the final month of pregnancy. The infant was born in a different state via cesarean section because of premature rupture of membranes. His length at birth was 47 cm (18.5 inches), his weight was 2.7 kg (6 lb), and his head circumference was 32 cm (12.5 inches). He stayed in the neonatal intensive care unit for 2 weeks for antibiotic therapy and management of jaundice. After discharge, he lived in a foster home for 2 weeks and was then transferred to the care of his maternal grandmother. The grandmother did not know if he experienced symptoms of drug withdrawal during the newborn period. Newborn hearing and metabolic test results also were unknown to the grandmother. Genetic tests had been ordered shortly after birth because of the infant's facial dysmorphism, but results were not available.

Family/Social/Developmental History

The infant lived with his grandmother and two aunts. His mother was receiving inpatient psychiatric care in another state. The grandmother reported that the child's

FIGURE 1. Lagophthalmos (the inability to close the eyelids completely) was noted bilaterally.



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language and motor development was slower than that of her other grandchildren. At 5 months of age, the infant was not rolling over and did not babble. He was taking a standard cow's milk–based formula by bottle but refused strained foods by spoon. Despite gastroesophageal reflux symptoms, his weight gain had been steady since birth. The family history was negative for relatives with abnormal head shapes and eye disorders.

Review of Systems

The review of systems was positive for abnormal head shape, proptosis (forward protrusion of the globe with respect to the orbit), stridor, a café-au-lait spot, and a leg hemangioma. He also had nocturnal lagophthalmos (partially open eyes when sleeping). He was an obligate mouth breather with stridorous respirations.

Physical Examination

The infant was alert and interactive. His length was 62 cm (24.4 inches; 10th percentile for age), his weight was 6.17 kg (13.5 lb; 10th percentile), and his head circumference was 40.9 cm (16 inches; 7th percentile). His head had a keel-shaped forehead secondary to synostosis, and a prominent metopic suture and coronal suture ridging were present (Figure 2). The nose shape was flattened, and the ala nasi appeared flared. He had hypertelorism with severe proptosis. The infant tracked well with his eyes. He was edentulous and breathing through his mouth. His chest examination was notable for referred upper airway noise, but breath sounds were clear. His heart sounds and spine were normal. There was no hepatosplenomegaly. The fourth and fifth toes of each foot curved medially. The infant appeared to be hypertonic. He had one café-au-lait spot on a wrist and a hemangioma on the right anterior lower leg.

At 5 months of age, this infant had obvious craniofacial deformities, including brachycephaly and orbital

FIGURE 2. Brachycephaly ("flat head syndrome"). The head is disproportionately wide compared with its depth.

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hypoplasia with associated proptosis, as well as developmental speech delay and obligate, stridorous mouth breathing. Based on these findings and experience with similar-appearing patients, we made a visual diagnosis of Crouzon syndrome. As primary care providers, we became the "medical home" for this infant and family. We managed the multispecialty team referrals, case coordination, and psychosocial support. In this article, we describe the extensive medical care provided to this child over a 7-year period. This coordination has allowed him to grow and develop in caring environments equipped to meet the needs of a medically fragile child.

Diagnostic Testing

We referred the patient to our university's genetics clinic for confirmation of the Crouzon syndrome diagnosis. The geneticist concurred that patient's facial features were consistent with Crouzon syndrome. Molecular tests to confirm this diagnosis could not be obtained because of insurance limitations. Because genetic testing was not necessary to initiate further treatment, we referred the patient to otolaryngology for a hearing test and for evaluation of his stridorous breathing.

Developmental Care

The craniofacial team members confirmed the grandmother's impression that the infant had delayed receptive and expressive language, fine motor delays, oral motor dysfunction, visual perceptive deficits, and abnormal posture. This assessment was based on direct observation. At subsequent well-child checks, Ages and Stages Questionnaires (Squires, Potter, & Bricker, 1990) were used for developmental screening. He was referred to the multidisciplinary craniofacial clinic where clinicians from neurosurgery, plastic surgery, oral and maxillofacial surgery, clinical social work,