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A Case of Saethre-Chotzen Syndrome

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Saethre-Chotzen syndrome was described independently by the Norwegian psychiatrist, Saethre,[1] and the German psychiatrist, Chotzen,[2] in the 1930s; since that time many cases have been reported, some using the terms acrocephalosyndactyly, type III, and craniooculodental syndrome. Clinically, the syndrome is characterized by premature closure of the cranial sutures, low-set hairline, nasal septum deviation, brachydactyly, and ptosis.[3] It is inherited as an autosomal dominant with complete penetrance and

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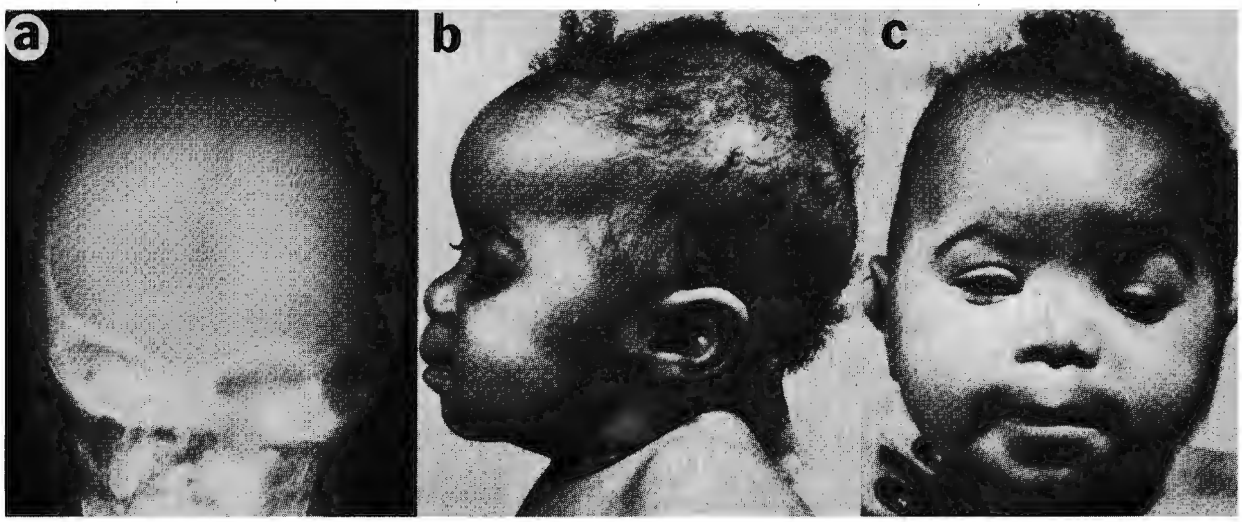

Figure—(*a*) Skull x-ray at 6 months reveals difference in orbit size and asymmetric shape of skull. (*b*) Left profile shows abnormal placement of ear and flat nasal bridge. (*c*) Patient at 6 months has left-sided ptosis, dystopia canthorum, and short palpebral fissures. Face shows characteristic asymmetries with left side being affected.

great variability in expression. Because of this variable in expressivity, the syndrome is difficult to diagnose in the less severe form without a positive family history.

Recently, an infant was admitted to the Medical College of Virginia newborn nursery who had many of the features of Saethre-Chotzen syndrome, as well as other findings which have rarely been reported in association with the syndrome.

Case Report

The patient is a black female infant who was born five weeks prematurely to a 17-year-old para 1, gravida 1 mother and a 23-year-old father. She weighed 1,830 gm and had a 27.5 cm head circumference at birth. At four weeks, she was transferred to the MCV Hospital with Group B-Beta streptococcal sepsis which was successfully treated with a ten-day course of penicillin and gentamicin. On admission, she was noted to have facial asymmetries, ptosis, hypertonia and a probable ventricular septal defect; she was referred for a genetics consultation.

Her family was evaluated for stigmata of Saethre-Chotzen syndrome. The child's father was unavailable for examination but was reported as having a face that was slightly asymmetrical with an under-developed left cranium and the left eye smaller than the right. These asymmetries were said to be more pronounced in childhood. The mother was examined and did not show any stigmata of the syndrome. No other family members were known to be affected.

The physical examination of the child revealed an acrocephalic skull with left-sided malar, frontal and anterior parietal bone hypoplasia, and microcephaly. Her face was asymmetric with left-sided ptosis; she had dystopia with generally small palpebral fissures, the left slit being shorter than the right. Her nasal bridge was flat, her nose had a broad tip and a long prominent philtrum, and her mouth was thin and down-slanting. The left ear was lower than the right and posteriorly rotated, but both ears were well formed. Her neck was short and displayed a decreased range of motion, though it appeared normal on x-rays. Her hips also displayed a 20 degree decrease in motion. She had two cafe-au-lait spots; one 2×3 cm on her back and one 2.5 cm in diameter in the groin area. There was a sacral dimple on her lower back. She had a grade IV/VI systolic murmur, probably caused by a ventricular septal defect.

Her extremities were slightly asymmetrical with the right arm and leg 0.5 cm longer than the left. She had two dimples on either side of each elbow and knee, a peculiar long longitudinal skin crease along both inner calves, and mild hallux valgus.

A dermatoglyphic analysis revealed an excess of whorls (six), one of which was a radial loop with a central pocket. She had an intermediate triradius. Both the a-b ridge count and the total finger ridge count were within normal limits.

X-rays of her skull and hips in the neonatal period were read as normal except for a normal variant of a lückenschädel deformity of the skull. Repeat x-rays of the skull at 6 months showed 1) cranial asymmetry with the left orbit smaller than the right, 2) coronal sutures that were patent but narrower than the other cranial sutures, and 3) a left temporal bone that was higher than the right (Figure-*a*). X-rays of the cervicothoracic spine at that time were normal. Chromosome analysis showed a normal female karyotype, and her serum calcium of 11 mg/100 ml was normal at 6 months.

A follow-up examination at age 8½ months showed all her physical measurements to be below the 3rd percentile with a length of 63 cm, weight of 13 lb 12 oz, and head circumference of 39.5 cm. She was generally hypertonic and

had poor head control. However, her motor milestones were not delayed; she was able to crawl and transfer objects from hand to hand by 8 months and sit without support at the age of 9 months.

From these physical and radiological findings the diagnosis of Saethre-Chotzen syndrome was made.

Discussion

The physical findings of 120 cases of Saethre-Chotzen syndrome were compared by O. A. Pantke et al.[4] In facial appearance the patients were strikingly similar to each other and to our patient. More than 75% had ptosis, deviated septum, acrocephaly, and low-set frontal hairline. The facial asymmetries and unusual cranial contours in the syndrome probably result from premature closure of the coronal or sphenobasilar sutures. This may also account for the tear duct stenosis and the mild mental retardation which are occasionally found. Skull x-rays like the ones in this case are characteristic, showing premature closure of the sutures as well as asymmetries in bone and orbit size. Hypertelorism and dystopia canthorum are often observed. Optic atrophy may be an associated finding.

The ears are frequently small and low-set, with folded pinnae. Conductive hearing loss is an occasional finding. The palate is either high-arched or clefted. Dental anomalies such as missing teeth and peg-shaped or anomalous maxillary lateral incisors are important in differientating this syndrome from the other acrocephalies.

The extremities may show mild syndactyly; brachydactyly is common. Clinodactyly and hallux valgus are sometimes noted.

The cardiac defect our patient exhibits is rarely found in Saethre-Chotzen syndrome; however, it was reported in Chotzen's original cases and in several individuals of another kindred.[5]

The multitude of associated findings in this syndrome can not be explained on the basis of premature closure of the sutures alone, though this does account for many of the facial features. Other malformations such as the cardiac defect, dental anomalies, and defects of the extremities indicate that one gene has varied effects on many organ systems.

Our case further documents the association of cardiac defects with Saethre-Chotzen syndrome while also demonstrating many of the characteristic findings. The child's father probably represents a very mild expression of this dominantly inherited condition, demonstrating the wide variability of expression seen in this disorder.

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