Auralcephalosyndactyly: a new craniosynostosis syndrome or a variant of the Saethre-Chotzen syndrome?

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SUMMARY A mother and son are reported with bilateral, symmetrical syndactyly of the third, fourth, and fifth toes, mild craniosynostosis of the coronary sutures, and small pinnae. The same combination of malformations was recently described as a new syndrome by Kurczynsky and Caspersen in a mother and her daughter.¹ In addition, in the present family, the mother had fusion of two cervical vertebrae and a partial duplication of the first metatarsal. The child had a bilateral cleft lip and palate. The question is raised whether these patients represent a new syndrome or a variant of the Saethre-Chotzen syndrome.

Saethre² and Chotzen³ each independently reported the association of mild acrocephaly and asymmetry of the skull associated with partial soft tissue syndactyly of the second and third fingers and the third and fourth toes. Several other reports have confirmed autosomal dominant inheritance and variable expression of this syndrome (for review see Temtamy and McKusick⁴).

Case report

The proband, a male, was the first child of unrelated parents. At birth a bilateral cleft lip and palate was noted, together with cutaneous syndactyly of the fourth and fifth toes (complete on the left side and partial on the right side). No problems were noted in the perinatal period. At the age of eight weeks he was referred with his parents for further diagnosis and genetic counselling. Length was 57.5 cm (50th centile), weight 4.4 kg (25th centile), and head circumference 38 cm (25th centile), all within normal limits, and psychomotor development was adequate for age. There was a bilateral total cleft palate; the cleft lip had already been repaired. He was brachycephalic with slight exophthalmos and upward slanting palpebral fissures (fig 1). Bilateral

preauricular pits and small ears were present. Examination of the chest, abdomen, and genitalia was normal. The fingers were relatively short and the feet showed the bilateral, symmetrical, cutaneous syndactyly already described and large toes (fig 2). Skull radiography showed a small anterior fossa and almost complete closure of the coronal sutures. Radiography of the extremities showed an abnormal, small, and curved fourth metatarsal and lack of ossification of the distal phalanges. Follow up at the age of four months showed normal psychomotor development and no evolution in the synostosis.

The mother had the same brachycephalic head shape with exophthalmos and ptosis (fig 3). She had small ears with incomplete development of the scaphoid fossa and small lobules. The uvula was short, but there was no cleft. Further clinical examination showed a short neck, short fingers, broad halluces, and asymmetrical, complete, cutaneous syndactyly of the toes (fourth and fifth toe on the left side and third, fourth, and fifth toes on

FIG 1 Proband at the age of four months, showing repaired lip, exophthalmia, and upward slanting palpebral fissures.

FIG 2 Feet of proband.

the right side) (fig 4a). Radiological examination indicated fusion of the second and third cervical vertebrae, an abnormally short and thin fourth metatarsal, partial duplication of the first metatarsal, and bifid terminal phalanges of both halluces (fig 4b).

Chromosomal analysis on peripheral lymphocyte cultures showed a normal G banded karyotype in

mother and son. Further family history is negative. The mother is the youngest of seven children, her father being 39 years and her mother 38 years at the time of her birth.

Discussion

Recently Kurczynsky and Casperson¹ described a mother and daughter with craniosynostosis and symmetrical syndactyly of the feet involving the fourth and fifth toes. In addition both had a short columella and small pinnae. The authors stated that the association of this particular syndactyly of the toes and the unusually shaped pinnae with craniosynostosis constitutes a new autosomal dominant acrocephalosyndactyly syndrome, and they designated it a new auralcephalosyndactyly syndrome.

The mother and son described in this paper present the same type of autosomal dominant acrocephalosyndactyly, but in addition there is a bilateral cleft lip and palate in the son, fusion of cervical vertebrae and broad halluces in the mother, and an abnormally shaped fourth metatarsal in both mother and son. These characteristics are described in the Saethre-Chotzen syndrome, another autosomal dominant acrocephalosyndactyly syndrome.⁴⁻⁶ The question may therefore be raised whether the family reported by Kurczynsky and Casperson¹ and the present family are examples of a new auralcephalosyndactyly syndrome or represent variable expression of the Saethre-Chotzen syndrome. Furthermore, another characteristic finding in the present family is the partial polydactyly in the

FIG 3 Face of the mother.

FIG 4a Feet of mother.

mother with broad first metatarsals and bifid distal phalanges of both halluces. These skeletal changes in combination with cutaneous syndactyly of the feet, abnormal auricles, and acrocephaly have been

described as an example of the Saethre-Chotzen syndrome⁷ and as the Robinow-Sorauf syndrome.⁸ The patients described in these two papers closely resemble the mother and son in this paper, except for the bilateral cleft lip and palate in the boy. Only the latter is reported in the Saethre-Chotzen, aural-cephalosyndactyly, and Robinow-Sorauf syndromes. We do not believe that they constitute separate entities, but more likely all these families represent different expression of the same dominant gene.

FIG 4b Radiograph of feet of mother. Note abnormal fourth metatarsal, partial duplication of first metatarsal, and bifid terminal phalanges of both halluces.

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