Complete situs inversus and broad thumbs and big toes with postaxial polydactyly

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Abstract

A healthy, non-consanguineous couple had a son with complete situs inversus viscerum (including dextrocardia but without other cardiac defects), broad thumbs and big toes, postaxial polydactyly, average intelligence and length proportion of the extremities, and a normal face. The common cause of these defects may have a role in the origin of sidedness and symmetry in morphogenesis.

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The development of sidedness and defects in lateralisation leading to the failure of normal symmetry in morphogenesis is an interesting basic question of organogenesis.¹ As far as we know, the combination of complete situs inversus, bilateral broad thumbs and big toes, and bilateral postaxial polydactyly has not been published previously. Thus, the purpose of this

case report is to contribute a new case to the topic of laterality.

Case report

A healthy couple visited our Genetic Counselling Clinic in 1982 with their son, born in 1974, who had a complex developmental disturbance. The parents were not consanguineous, their family history was negative, and they had no other child. The proband was born at term after an uncomplicated pregnancy weighing 3200 g. His broad thumbs and big toes, bilateral postaxial skin tag polydactyly of the hands, and unilateral polydactyly of the left foot were noted immediately and his extra digits were removed. His situs inversus viscerum (transposition of the liver, one spleen of normal size, and large intestines, the right testis hanging lower than the left unlike normal) with dextrocardia was detected later. The proband returned in 1996 to clarify the recurrence risk of his defect for his planned offspring. He had a normal height (175 cm) with average intelligence and weight (70 kg) without any disproportion of the extremities or facial dysmorphism. His dextrocardia with total situs inversus viscerum was confirmed, but other cardiological defects were excluded by echocardiography. His hands with broad thumbs and simian creases (fig 1) and his feet with broad big toes (fig 2) showed a characteristic radiographic pattern (figs 3 and 4). Both thumbs had broad metacarpals and phalanges and the terminal phalanges were shorter. The second and third fingers showed a slight ulnar deviation and the third metacarpal was a bit broader. The fourth and fifth metacarpals were shorter with broader proximal epiphyses. All but the first terminal phalanges were normal. The first and fifth metacarpals had a concave angulation. In the carpal bones there was an

Figure 1 Hands of the proband.

Figure 2 Feet of the proband.

Figure 3 X ray of the hands.

Figure 4 X ray of the feet.

extra os parastyloides. Both big toes had broad metatarsal and phalangeal bones with shorter terminal phalanges. The latter was also seen in the second and third toes. The fourth metacarpal bones were shorter. There was a joint between the medial and lateral parts of the os naviculare in both feet and he had bilateral, mild hallux valgus.

Discussion

The developmental disturbance described comprises three defects: (1) well formed skin tag postaxial polydactyly, probably type B (that is, pedunculated postminimi) which was removed. However, types A and B postaxial polydactyly may be genetically identical because these two forms have been found in the same families.^{2,3} (2) Broad thumbs and big toes predominated in the complex bone developmental disturbance of the hands and feet. (3) Complete situs inversus. There is evidence both in man⁴ and mouse⁵ that complete situs inversus and heterotaxy can represent a spec-

trum of defects and the laterality sequence may include polysplenia,⁶ asplenia-hypoplastic spleen (Ivemark syndrome),⁷ and lung and cardiovascular malformations.^{8,9} Our proband does not have these defects (except dextrocardia as part of situs inversus, so the pattern seems to be an independent entity). Linkage of X linked laterality sequence to Xq24-q27.1 has been reported,¹⁰ while homozygous mutations in the connexin43 gap junction gene have been found in patients with laterality defects.¹¹

In the differential diagnosis, Kartagener's syndrome was unlikely because the proband had no defects of the cilia (nasal cilia were normal) and its consequences (sinusitis, bronchiectasia, etc). Sperm tails were not examined as the patient was non-compliant for religious reasons. Ellis-van Creveld syndrome was excluded because he had no short distal extremities, nail hypoplasia, cardiac defect, or reduced height. Rubinstein-Taybi syndrome was also excluded because of the lack of mental retardation and characteristic face, and Greig cephalopolysyndactyly syndrome because of the lack of syndactyly and peculiar skull shape.

In conclusion, the chance combination of the three developmental defects in the same person is unlikely, so a common cause can be expected. This cause may have a general importance in the origin of sidedness and symmetry in morphogenesis.¹²

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