Heptacarpo-Octatarso-Dactyly Combined with Multiple Malformations

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Abstract. A case of heptacarpo-octatarso-dactyly combined with cheilo-gnatho-palato-schisis, hypertelorism, macroglossia, complex malformations of heart and great vessels, horse-shoe-kidney, micro-penis, and penis palma is described. To our knowledge, this syndrome has not yet been described in the literature. Some of the features of the case reported overlap with syndromes such as Grauhan syndrome and Meckel syndrome.

Key words: Heptacarpo-octatarso-dactyly - Hydrocephalus - Complex malformations of heart and great vessels - Horseshoe-kidney - Micro-penis

Polydactyly very frequently is accompanied by other malformations and at least 13 different syndromes (Panum 1878; Reber 1967/68; Temtamy and Mckusick 1969, 1978; Kelikian 1974) have been described in which it is a leading feature. However, none of these syndromes seems to encompass the case reported here.

Case Report

The baby was born to a 28-year-old German gravida 1 and a 21-year-old Yugoslavian man. There was no consanguinity and no exposure to any infections, radiation or drugs. Pregnancy was normal although at the end of it hydramnios developed, accompanied by a weight gain of 17 kg. Delivery was spontaneous. The child was referred to the neonatal intensive care unit of the Center of Pediatrics because of his poor general condition and his malformations.

The baby's weight was 3,200 g (25th percentile), length 54 cm (90th percentile), fronto-occipital circumference 39 cm (90th percentile) according to a modified Farr-Score for a newborn of 38 weeks of gestational age and local growth chart. Physical examination revealed a critical condition with livid grey skin, tachypnoea, and a systolic heart murmur. Repeated convulsions were seen. The normal newborn reflexes and reactions could not be elicited and the child died after 17 h.

Multiple Malformations

Clinical and Postmortem Investigation. Cheilo-gnatho-palatoschisis; hypertelorism; macroglossia; macrocephaly (Fig. 1) with internal hydrocephalus including all four ventricles; single hazel-nut sized cysts within the ependyma of the lateral ventricles; polgyria; leptomeningosis fibrosa at the base of the brain; accessory superior lobe of the right lung with a high origin of the bronchus belonging to it.

In addition there was hepatosplenomegaly; horseshoe-kidney; micro-penis and penis palma (Fig. 2). Polydactyly was as follows: Six fingers and two thumbs on each hand (Fig. 3). Seven toes with duplication of the first toe of the right foot and eight toes with triple phalanges, fused first toe with a small accessory toe on the left foot (Fig. 4).

Cardiac Section. Complex malformations of the cardiovascular system: There was a complete persistent atroventricular canal. Mitral and tricuspid valve showed a common valvular ring with a common anterior leaflet. Ventricular arterial connection was normal. The main pulmonary trunk continued in a left pulmonary artery and a patent ductus arteriosus. The right pulmonary artery arose as the first vessel from the ascending aorta proximate to the ascending aorta proximate to the normal origin of the right subclavian and aortic arteries.

X-ray Examination. The right hand revealed a heptadactyly of the phalanges I–VII with soft tissue fusion of the thumbs I–II and a hexadactyly of the metacarpals I–VI with an osseous base defect of V. The left hand showed heptadactyly of the phalanges I–VII and a hexadactyly of the metacarpals I–VI with a proximal osseous fusion of the metacarpals I–II. The right foot exhibited octodactyly of the distal phalanges I–VIII including soft tissue fusion of the great toes II–III, and heptadactyly of the metatarsals I–VII (Fig. 5).

The left foot revealed octodactyly of the distal phalanges I–VIII including soft tissue fusion of the great toes II–IV, hexadactyly of the proximal phalanges I–VI, and hexadactyly of the metatarsals I–VI.
Chromosomes from the patient were prepared from a peripheral blood culture and from fibroblasts in culture. The analysis of the chromosomes with Q-, G-, and C-banding showed a normal male karyotype: 46,XY. Both parents showed a normal karyotype when chromosomes were prepared from peripheral blood.

Dermatoglyphic Patterns. Altogether, 14 whirls at the fingertips of the right and left hands; simian line on the right hand; an additional two-finger-grove in the left palm; altogether at the tips of the toes at least 10 arches. Because of postmortem maceration the palmar and plantar dermatoglyphics could not be evaluated. Dermatoglyphic pattern of the mother: 5 arches, 3 loops, and 2 whorls; a distinct ulnar shift of an axial triradius on the right and left palms; transition form of simian line on the left. Dermatoglyphs of the father: 6 whorls and 4 loops at the fingertips; remarkable large loop pattern on both hypothenar areas with additional T' and T".

Discussion

Our patient appears to present a syndrome which has not yet been described in the literature, although some of the features overlap with known syndromes, such as the Grauham syndrome characterized by polydactyly, cheilo-palato-schisis, urogenital malformations (Leiber and Olbrich 1972) and the Meckel syndrome also called Gruber syndrome (Gruber 1934) characterized.