Case reports

The acrocallosal syndrome in a Turkish boy

Memnune Yüksel, Mine Çaliskan, Gönül Oğur, Meral Özmen, Gülderen Dolunay, Selçuk Apak

Abstract

A 6 month old Turkish boy with the acrocallosal syndrome is reported. The patient, born to consanguineous, healthy parents, presented with macrocephaly, a prominent forehead, hypertelorism, polydactyly of the fingers and toes, severe motor and mental retardation, hypotonia, and absence of the corpus callosum. The mode of inheritance is discussed and our case is compared with previously reported cases of the syndrome.

The acrocallosal syndrome was first described by Schinzel¹ in 1979. We report a male infant born to consanguineous parents with similar craniofacial dysmorphism, postaxial polydactyly of the hands and feet, severe motor and mental retardation, hypotonia, cyanotic spells, and absence of the corpus callosum.

Case report

The patient was first examined at 40 days of age because of polydactyly and dysmorphic features. He was the second child of healthy, young, Turkish parents who were first cousins. The first pregnancy had ended in spontaneous abortion during the first trimester.

Delivery was uneventful, birth weight was 3500 g, length 50 cm, and occipitofrontal head circumference 36 cm. Neonatally he was placed in an incubator because of several brief cyanotic spells. When first examined at 40 days of age the patient showed the following dysmorphic features (fig 1): large anterior fontanelle, high and prominent forehead, hyper-

telorism (ICD 35 mm, >97th centile), downward slanting palpebral fissures, epicanthic folds, a broad nasal bridge, short nose and philtrum, prominent ears, and an open mouth with protruding tongue. The hands showed bilateral postaxial polydactyly (fig 2). Scars from the postaxial supernumerary toes that were removed at birth could be seen. There was a right undescended testis. Neurological examination showed marked generalised hypotonia.

Cranial ultrasound and CT scan (fig 3) indicated absence of the corpus callosum. EEG showed asynchrony with abnormal background activity. X rays of

Figure 1 The patient at 4½ months of age.

Figure 2 Hands of the patient showing bilateral postaxial polydactyly.

Division of Genetics, Institute of Child Health, University of Istanbul, Çapa-Istanbul, Turkey.

M Yüksel, M Çaliskan, G Oğur, S Apak

Department of Paediatrics, University of Istanbul, Turkey.

M Özmen, G Dolunay

Correspondence to Professor Yüksel.

Received for publication 13 March 1989.

Revised version accepted for publication 20 June 1989.

the skeleton were normal except for postaxial polydactyly of the hands (fig 4); the supernumerary rays originated at the bases of the proximal fifth phalanges and contained two phalanges. Ophthalmological and cardiological examinations including echocardiography were normal as were GTG banded chromosomes (46,XY). The patient was followed up to 6 months of age. During this period he was admitted to hospital several times for cyanotic spells, hyperthermia, and respiratory tract infections.

At his last admittance to hospital with bronchopneumonia at 4½ months his weight was 6 kg (10th to 25th centile), height 62 cm (10th to 25th centile), and head circumference 43 cm (50th to 75th centile). Mental and motor retardation were obvious, with marked hypotonia and poor social contact. He stayed in hospital for around one and a half months. The family informed us later that the baby had died 20

Major clinical features of our patient and previously reported cases

Clinical findings	Published cases (n=12)	Our case
Hypoplastic or absent corpus callosum	11/12	+
Postaxial polydactyly of hands	11/12	+
Postaxial polydactyly of feet	7/12	+
Duplication of phalanges of big toe	8/12	—
Craniofacial dysmorphism	12/12	+
Severe mental retardation	11/12	+
Cryptorchidism/hypospadias	2/6	+
Inguinal hernias	8/12	—
Cyanotic spells	3/12	+
Frequent infections	5/12	+
Hypotonia	9/12	+

days after his discharge from hospital. No necropsy was performed.

Discussion

Our patient increases the number of reported cases with the acrocallosal syndrome to 13.²⁻⁸ A comparison of the main features of the previous cases with our patient's is shown in the table. Total or partial absence of the corpus callosum, craniofacial dysmorphism, severe mental retardation, and postaxial polydactyly constitute the most consistent clinical features. All reported cases of the acrocallosal syndrome, apart from two families,^{7,8} were sporadic. It was previously assumed that the acrocallosal syndrome is caused by a fresh mutation of a dominant gene. Parental consanguinity in this and other reported cases⁴ and two affected cases in two families (sibs and first cousins) strongly suggest autosomal recessive inheritance. However, the rate of consanguinity in Turkey is among the highest (21.2%) in western countries⁹ and thus consanguinity is not such a strong argument for a recessive gene as it would be in a population with a low consanguinity rate.

Figure 3 CT scan showing absence of the corpus callosum.

Figure 4 X ray of left hand showing postaxial polydactyly and normal thumb.

- Schinzel A. Postaxial polydactyly, hallux duplication, absence of the corpus callosum, macrencephaly and severe mental retardation: a new syndrome? *Helv Paediatr Acta* 1979;34:141-6.
- Legius E, Fryns JP, Casaer P. Schinzel acrocallosal syndrome: a variant example of the Greig syndrome? *Ann Genet (Paris)* 1985;28:239-40.
- Nelson MM, Thompson AJ. The acrocallosal syndrome. *Am J Med Genet* 1982;12:195-9.
- Philip N, Apicella N, Lassman J. The acrocallosal syndrome. *Eur J Pediatr* 1988;147:206-8.
- Sanchis A, Cervero L, Martinez A. Duplication of hands and feet, multiple joint dislocations, absence of corpus callosum and hypoarrhythmia: acrocallosal syndrome? *Am J Med Genet* 1985;20:123-30.
- Schinzel A, Schmid W. Hallux duplication, postaxial polydactyly, absence of the corpus callosum, severe mental retardation and additional anomalies in two unrelated patients: a new syndrome. *Am J Med Genet* 1980;6:241-9.
- Schinzel A, Kaufman U. The acrocallosal syndrome in sisters. *Clin Genet* 1986;30:399-405.
- Schinzel A. The acrocallosal syndrome in first cousins; widening of the spectrum of clinical features and further support for autosomal recessive inheritance. *J Med Genet* 1988;25:332-6.
- Başaran N, Şaylı BS, Başaran A. Consanguineous marriages in the Turkish population. *Clin Genet* 1988;34:339-41.