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18p− syndrome resulting from translocation (13q;18q) in a mildly affected adult male[1]

SUMMARY The patient was a 27-year-old male with short stature, borderline mental deficiency, strabismus, and a short fourth metacarpal. His karyotype showed deletion of the short arm of a chromosome 18 as the result of *de novo* fusion centric translocation between chromosomes 13 and 18 (45,XY,−13,−18,+t(13;18) (13qter→cen→18qter).

Since the first description of a case with deletion of the short arm of chromosome 18 (18p−) by de Grouchy *et al.* (1963), over 80 cases have been reported. The phenotypic expression of 18p− cases is variable: the common features are growth and mental deficiency, hypertelorism, epicanthic folds, and large, protruding, and low set ears (Faust *et al.*, 1976). The association of arhinencephaly and cebocephaly with 18p− is also known (Lurie and Lazjuk, 1972). Many of the features of Turner's syndrome have also been reported frequently in these cases, consisting of short and webbed neck, lymphoedema at birth, and shield or funnel chest with widely set nipples (de Grouchy, 1969; Lurie and Lazjuk, 1972).

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A phenotype of Goldenhar's syndrome associated with 18p− has also been described (Buffoni *et al.*, 1976).

Variability in the phenotypic expression of 18p− is further illustrated by our patient, who exhibited only mild dysmorphic features and borderline intellectual capacity.

Case report

This case was a 27-year-old man referred because of cognitive and adaptive deficiency. He was the product of an uncomplicated term pregnancy, with a birthweight of 3600 g. The mother and father were 22 and 24 years old, respectively, at the time of his birth. Recorded length at 2 weeks of age was 52·5 cm. He was first evaluated at age 2 years 3 months for short stature and language delay, but no diagnosis was made.

The patient was in classes for the educable mentally retarded throughout his school years. He has had alternating strabismus since childhood, and a right inguinal herniorrhaphy was done at the age of 18. A cutaneous basal cell carcinoma of the right arm was removed recently at the age of 27.

The father's height was 180 cm and the mother's 166 cm. The paternal grandmother's height was 145 cm, and there was no history of short stature in the mother's family. The mother had had a total hysterectomy at the age of 42 years for carcinoma of the uterus, and her father had lung carcinoma. There was one sib, a girl, who died at the age of 3½ because of leukaemia.

Physical examination at age 27 showed the following. Height was 152·5 cm, weight 61·7 kg, and head circumference 56 cm. Arm span was 148·7 cm with an upper/lower segment ratio of 0·84. His blood pressure was 110/80. Unusual craniofacial features were noted as follows (Fig. 1): alternating internal strabismus, posteriorly rotated ears, and a rather small mandible. The neck was short and broad with normal range of motion. The teeth were carious but not malformed, and hair and nails were normal. Apart from one café-au-lait spot below the left knee, the skin was normal. The hands and feet were broad and short, and the left fourth metacarpal was short. Except for a speech articulation problem, the neurological examination was normal. Dermatoglyphs showed both palmar axial triradii in the t″ position and a digital pattern of 10 ulnar loops. The mother had a digital pattern of 10 ulnar loops, the left palmar axial triradius in the t′ position, and the right in the t position. The father's dermatoglyphs consisted of 8 ulnar loops, 1 radial loop, and 1 whorl, with both palmar axial triradii in the t position.

Psychological evaluation showed the patient's

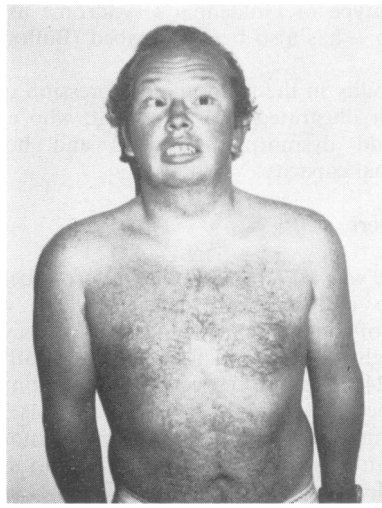

Fig. 1 *Proband at 17 years of age. Note the esotropia and the short, broad neck.*

cognitive and social-adaptive function to be at the borderline retardation level, with mild to moderate perceptual-motor dysfunction, mild difficulty with impulse control, and no psychiatric disturbance. On the Wechsler Adult Intelligence Scale he had a verbal IQ of 81, performance IQ of 69, and a full scale IQ of 75.

The patient held a job as a janitor and was driving himself to work at the time of evaluation.

In view of the mild mental retardation, short stature, and one short fourth metacarpal, pseudo/pseudopseudohypoparathyroidism was considered. Further examinations, consisting of *x*-ray of the wrists, hands, and cervical spine, as well as serum calcium, phosphorus, growth hormone, prolactin, and T4, were all normal. Urine metabolic screening was also normal.

CYTOGENETIC STUDIES

Chromosome analysis was performed on peripheral blood lymphocytes with Giemsa (Seabright, 1971), C-banding (Sumner, 1972), and silver stain (Bloom and Goodpasture, 1976). Giemsa banding showed a centric fusion translocation between the arms of chromosomes 13 and 18, 45,XY,−13,−18,+t (13;18) (13qter→cen→18qter). The C-banding showed only a single heterochromatin region. The silver stain showed no evidence of a nucleolar organising region (Fig. 2). Giemsa banding of the patient's skin fibroblasts also showed the same chromosomal change. The parents' karyotypes were normal. Sequential quinacrine and silver staining was done on the patient's and his parents' chromosomes 13 in an attempt to determine the parental origin of the chromosome rearrangement. The normal chromosome 13 of the patient had dark silver staining and

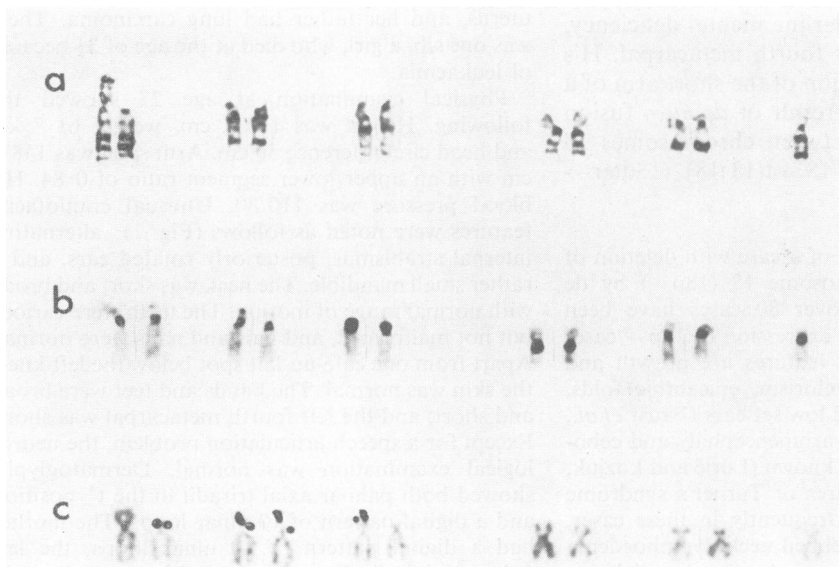

Fig. 2 *Partial karyotype (D and E group) on peripheral lymphocytes of proband with (a) Giemsa banding, (b) C-banding, (c) silver stain.*

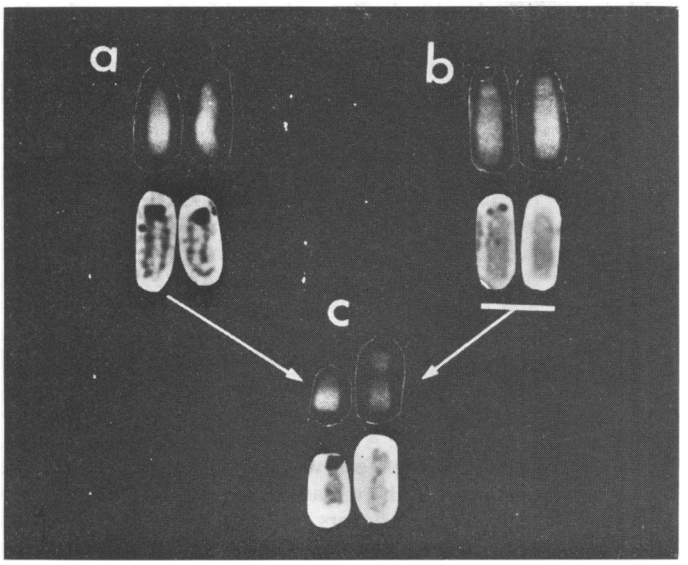

Fig. 3 *Sequential quinacrine and silver stain of chromosomes 13 of (a) the father, (b) the mother, and (c) the patient, suggests the translocated chromosome is maternal in origin (see text).*

fluorescent satellites which matched one of the father's chromosomes 13 (Fig. 3).

Discussion

Of more than 80 cases with 18p− (Faust *et al.*, 1976), at least 12 cases were the result of a translocation involving chromosome 18 and one of the acrocentric chromosomes. Almost half of these cases had a carrier parent, and the remainder were *de novo* cases (Lurie and Lazjuk, 1972). Deletion of the short arm of 18 in the present case was the result of a *de novo* centric fusion translocation between the long arms of chromosomes 13 and 18. The C-banding showed only one heterochromatin block, and the silver stain did not show evidence of the nucleolar organising region, suggesting that a substantial part of the short arms of chromosomes 13 and 18 was deleted.

The patient reported here is the second such case detected in our laboratory. In contrast to the first case of a 26-month-old male with congenital heart disease, IgA deficiency, short stature, and more severe language deficiency (Funderburk *et al.*, 1977), our present case is only mildly affected. The present case, therefore, supports the observation that, although the phenotype of 18p− depends upon the loss of the short arm of chromosome 18, there is no correlation between the extent of the deficiency of the short arm and the expression of malformations (Faust *et al.*, 1976).

It is interesting to note that the patient, his sister, his mother, and his maternal grandfather all had neoplastic disease. Furthermore, sequential quinacrine and silver stain showed that the translocated chromosome in the patient originated from the mother. This raises the speculation of the possible relationship between the maternal chromosome constitution and neoplastic disease.

Our patient had some features suggestive of pseudo/pseudopseudohypoparathyroidism: short stature, mental deficiency, and one short fourth metacarpal. Our patient, therefore, emphasises that chromosome analysis may be indicated in patients with clinical features of a non-chromosomal syndrome, as well as in patients with only mild dysmorphic features and borderline intellectual capacity.

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Brachydactyly and polydactyly with dermal ridge dissociation and ridge hypoplasia

SUMMARY A child with brachymesophalangy and postaxial postminimal polydactyly was found also to have dermal ridge dissociation and ridge hypoplasia. She was the second child of unaffected, unrelated parents and was born after a normal pregnancy and delivery. No previous report of a similar combination has been traced.

Brachydactyly may either occur as an isolated developmental defect, usually inherited in an autosomal dominant manner, or as part of a more complex malformation not confined to the extremities. The classification of isolated brachydactyly is based on that of Bell (1951), who recognised three types with brachymesophalangy (shortening of middle phalanges) alone, types A1, A2, and A3; three further forms of brachyphalangy, types B, C, and D (stub thumbs); and a form with brachymetacarpy and brachymetatarsy, type E. In 1970, Edwards and Gale described a mixed type combining brachymesophalangy, brachymetacarpy, brachymetatarsy, and camptodactyly. More recently, Christian *et al.* (1972) reported a new form of preaxial brachydactyly with short thumbs and first toes angulated at the metacarpophalangeal or metatarsophalangeal joint. Bilginturan *et al.* (1973) described a new form of combined brachyphalangy, brachymetacarpy, and brachymetatarsy associated with hypertension. Other developmental disorders associated with brachydactyly that have been reported include joint dysplasia (Liebenberg, 1973), and cerebellar ataxia with nystagmus (Biemond, 1934). Since Bell's account, two new types of brachymesophalangy have been described, that of Temtamy (1966), type A4, and that of Bass (1968), type A5. Schott (1978) has also recently published an account of a family with a new form of type B brachydactyly (shortening of terminal and, to a lesser extent, middle phalanges), and Sillence (1978) has indicated further heterogeneity in type A1 brachydactyly. In Schott's family, hands, but not feet, were involved, the thumbs were spared, and there was an associated nail dysplasia of the affected fingers. The findings on the hands and feet of the five members of a kindred seen by Sillence were consistent with type A1 brachydactyly, but they also had tall normal stature, scoliosis, and club feet. None of these different types of brachydactyly involved polydactyly, dermal ridge dissociation, and ridge hypoplasia as was seen in the sporadic case reported here.

Case report

The patient was a girl born on 18.7.71. She was born at term after a normal pregnancy and delivery, with a birthweight of 3·97 kg. Her mother was 27 years old and her father 32 years at the time of her birth. A normal elder sister was born in June 1970. The patient was noted at birth to have bilateral small fleshy fingers with nails, attached by a narrow piece of skin to the proximal phalangeal region of the outer border of the fifth fingers. There was a similar postaxial toe on the right foot but not on the left. All three extra digits were removed shortly after birth.

She was seen, with her parents and sister, in 1973 for an opinion relating to her possible acceptance for immigration abroad. At that time, direct examination of her parents and sister showed no abnormality of hands or fingers and no history could be obtained of such abnormality in any further member of the family. There was no parental consanguinity (Fig. 1).

On examination, the patient showed small scars where her extra digits had been removed. The second to fifth fingers of both hands were slightly short, but her thumbs were of normal length (Fig. 2). There was some limitation of flexion of the distal joint of the right thumb. There was no further abnormality of the limbs other than in the digits and she had