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Partial trisomy 6p with karyotype 46,XY,der(22), t(6;22)(p22;q13)mat

SUMMARY A case of partial trisomy 6p is reported with a review of the various characteristics of this syndrome.

Case report

The propositus was the second child of unrelated healthy parents, born only 10 days before term after an uneventful pregnancy. The father was then 30, the mother 25, and the elder healthy brother 2. The mother had had a spontaneous abortion at 2\(\frac{1}{2}\) months before having her two sons. At birth the propositus weighed 1800 g. He spent the first 3 months of his life in 2 different paediatric departments where he had feeding difficulties and failed to thrive. X-rays taken during that period showed normal bones in the skull, chest, and pelvis. Blood urea, glucose, and cholesterol, as well as serum immunoglobulin levels, phenylalanine, and sweat test, were normal. There were no reducing substances in the urine.

Feeding problems continued at home. He vomited frequently and could not swallow solid food; at 13 months he still only took milk. He was admitted to our unit with a persistent cough due to pertussis. Clinical examination showed severe psychomotor and growth retardation, with height, weight, and head circumference below the 3rd centile. The child had an odd facies (Fig. 1a) with slight micrognathia and low-set, large, simple ears with a very thin helix (scapha) without antitragus and posterior branch of the anthelix (Fig. 1b). He had extremely long eyelashes, eyes which were always closed, bilateral cataracts, a long philtrum, no definite border to the vermillion, no teeth, an enlarged abdomen, an omphalocele, a deep sacral dimple, normal genitals, and normal hands and feet without abnormal creases. No heart defect could be detected. His skin was dry and his hair thin and sparse. A day after admission he had convulsions without fever. Multiple cultures for bacteria in blood, urine, and spinal cord fluid were negative. The electrolytes showed low sodium, low chloride, and high potassium. Proteinuria was also significant. The child died within 10 days with symptoms of encephalitis due to pertussis. The parents did not allow necropsy.

Fig. 1 (a) The propositus at 13 months. (b) His ear is shown in inset.
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Fig. 2 The mother’s karyotype: 46,XX,15ps+,-t(6;22)(p22;q13).

Shortly before death blood had been taken for a cytogenetic evaluation.

Cytogenetics

Conventional harvesting and analysis of lymphocyte metaphases showed 46 chromosomes in all cells. G-banding revealed enlarged satellites on a chromosome 15, and an abnormal 22 with an extra dark band at the distal end of its elongated long arms.

The father had a normal karyotype, with large satellites on a 14 and on a 15. The mother also had large satellites on a 15, as well as a translocation involving a chromosome 6 and a chromosome 22, with points of exchange at bands 6p22 and 22q13 (Fig. 2). Her karyotype was thus 46,XX,15ps+-t(6;22)(p22;q13).

The patient had inherited her abnormal 22, as well as her normal 6, and he was trisomic for the segment 6pter to 6p23 and part of 6p22. His karyotype was thus 46,XY,15ps+-der(22),t(6;22)(p22;q13)mat. Other family members declined invitations to be karyotyped. No other abnormal child was known among the mother’s relatives.

Discussion

At least 8 other cases of partial trisomy 6p are known (Gouw et al., 1973; Breuning et al., 1977; Kjessler, 1977) associated with translocations onto chromosomes 2, 15, 18, and 20. Some of these children also had a partial monosomy for the other chromosomes involved in their translocations, but it is possible to recognise the characteristics that our patient shared with most of them, and that seem to form a distinct syndrome. These were: low birthweight after a normal gestation, severe psychomotor retardation and failure to thrive, large simple ears with hypoplastic antitragus and posterior branch of the anthelix, closed eyes or eyelid ptosis or phimosis, long philtrum, small pointed chin, sacral dimple, proteinuria, and death before 3 years of age. Heart defects are common but were not found in our patient.

A small deletion of the long arm of chromosome 22 might also be responsible for some of our patient’s characteristics. Indeed, the extremely long eyelashes, not reported in the other cases of trisomy 6p, might belong to the ring 22 syndrome (Hunter et al., 1977), as might a ‘simple vermilion border with long, flat
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 philtrum’ and high serum potassium (Palmer et al., 1977).

The technical assistance of Miss E. Pandelia is gratefully acknowledged.

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References


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Osteosarcoma in a patient with Hutchinson-Gilford progeria¹

SUMMARY A 13-year-old female with Hutchinson-Gilford progeria, who developed an osteosarcoma of the right chest wall, is reported. This is the first reported association of a malignant neoplasm with this syndrome.

Sir Jonathan Hutchinson first described a patient with ‘congenital absence of hair and its appendages’ in 1886. A second patient was reported in 1895 by Hutchinson. Gilford re-examined these patients and in 1904 described the pathological changes of the disease and termed these clinical findings ‘progeria’. Many patients have been reported in the intervening 75 years (DeBusk, 1972). The present report is the first example of a malignancy diagnosed in a patient with progeria.

¹Supported by a grant for a Genetics Study Center (PHS 10H 82 0004) and by a Mental Retardation Training Program no. 920 for the National Institutes of Health.

Case report

A 13-year-old white female (Fig. 1) presented with a dry cough and stabbing right anterior superior chest pain. She had a month long history of increasing fatigue. The early childhood history of this patient has been previously reported, along with in vitro replicative studies of her fibroblasts, by Martin et al. (1970). The typical clinical features of progeria were noted at approximately 2 years of age and were even more dramatic at the present admission. No pubertal secondary sex changes had occurred. In spite of the loss of vision in one eye because of infectious complications following strabismus surgery at age 2½ years the patient has been a ‘straight A’ student.

She was 103 cm tall with a weight of 13 kg. The right chest was dull to percussion with decreased breath sounds and scattered rales. Chest x-ray (Fig. 2a) revealed a large extra-pleural mass with loss of integrity of the 7th, 8th, and 9th ribs. A percutaneous biopsy was performed with a histological diagnosis of chondrosarcoma. No metastases were identified. An en-bloc resection of a portion of the right chest wall and ribs 5 to 9 was performed (Fig. 2b). The chest wall was closed with Marlex, and considering the magnitude of the procedure the patient had an uncomplicated postoperative course with ventilatory support for only 24 hours. Grossly, the resected tumour was not encapsulated. It consisted of the major portion of ribs 5 to 9 and their accompanying muscles, except the latissimus dorsi. The tumour mass was composed of fibrous yellow-grey connective tissue with local calcifications. The margins of resection were free of

Fig. 1 Patient at 13 years; she is wearing a wig.
Partial trisomy 6p with karyotype 46,XY,der(22), t(6;22)(p22;q13)mat.

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*J Med Genet* 1978 15: 479-481
doi: 10.1136/jmg.15.6.479

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