Partial Trisomy of 13(pter→q12) due to 47,XY,+der(13),t(13;22)(q12;q13)mat

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Summary. A 36-month-old boy presented with short stature, short neck, shield-shaped chest, and mental retardation. Chromosome analysis showed trisomy for the short arm and the proximal portion of the long arm of chromosome 13 [47,XY,+der(13),t(13;22)(q12;q13)mat]. The patient’s mother has a balanced translocation between the long arms of chromosomes 13 and 22 [46,XX,t(13;22)(q12;q13)]. The patient’s neutrophils showed an elevated number of nuclear projections and his fetal hemoglobin level was undetectable.

Introduction

Numerous cases of full trisomy 13 syndrome have been reported; partial trisomy for the long arm of chromosome 13, on the other hand, has been described in only about two dozen cases, ten of whom represent trisomy of the proximal portion of the long arm of chromosome 13. Whereas clinical features of the full trisomy 13 are well established, those of trisomy for the proximal portion of the long arm of chromosome 13 are not well defined.

We describe a patient who presented with short stature, retardation, and few minor anomalies. His karyotype showed an extra acrocentric chromosome, which was identified as the proximal segment of the long arm of chromosome 13 and its short arm.

Case Report

JD was referred at the age of 36 months for evaluation of delayed development. He was the product of a full-term uncomplicated pregnancy to an 18-year-old primigravida mother and 19-year-old father. His birth weight was 2.8 kg; the birth length is not known. He seemed to do well
until he was about 1 year old, when his mother became concerned about his small size and slow development. One afebrile seizure occurred at age 2; no cause was found and seizures have not recurred. Evaluation at the age of 37 months showed an overall developmental age of 20 months, which gave him a developmental quotient of 54. His size was that of an 18-month-old child in height (82.5 cm) and weight (10.8 kg), while his head circumference was that of a 10-month-old child (45.5 cm). Right esotropia was still present, even though he had had corrective surgery for this problem earlier. He had bilateral epicanthal folds and a mild antimongoloid slant of the palpebral fissures, with an inner canthal distance of 3 cm and outer canthal distance of 8 cm. His ears were not low-set and were normal in configuration. He also had a slightly elevated palate, a short neck, and a shield-shaped chest (Fig. 1). No abnormalities of the lungs, heart, abdomen, and genitalia were found. His muscle tone was normal. He had no speech and was hyperactive. A hearing test revealed no apparent hearing loss.

Dermatoglyphics showed both palmar axial triradii in the t' position, four digital ulnar loops and one arch on the right and three arches with two ulnar loops on the left side. Dermatoglyphics of the patient’s mother showed bilateral t’axial palmar triradii, and on the digits three arches, one radial loop, and six ulnar loops.

The patient’s parents are not related, the mother is 157.5 cm tall and the father is 172.5 cm tall. The half-brother of the patient’s mother is said to be slow (he has not been evaluated by us). The patient’s mother is American-Indian and his father is of Mexican descent. The family history is otherwise unremarkable for short stature or birth defects.