

## Dysmorphology report

# Unknown syndrome. A possible new X linked retardation syndrome: dysmorphic facies, microcephaly, hypotonia, and small genitalia

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### Abstract

The proband, the first child of unrelated parents, was noted in infancy to have microcephaly, developmental delay, dysmorphic facies, hypotonia, a small penis with cryptorchidism, and a fixed flexion deformity of his left index finger. His maternal uncle is severely retarded and has similar dysmorphic facies.

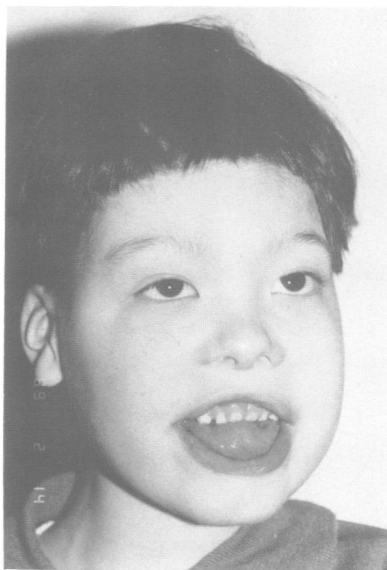
### Case report

The proband was born after a term pregnancy during which little fetal movement was felt. Delivery was vaginal and the neonatal period uncomplicated.

Over the next few months, he was noted to have developmental delay along with the following anomalies: scaphocephaly, medial eyebrow flare, short nose, thick lips, carp mouth, flat nasal bridge, hypodontia, fixed flexion deformity of the left index finger, cryptorchidism, small penis, and a prominent antihelix to the ear (figure).

Linear growth has been normal (35th centile for gestational age), but the head circumference is growing along a line parallel to but below the 3rd centile (46.6 cm at the age of 3 years 4 months). He is severely retarded and has no words at the age of 6. He is hypotonic and areflexic.

A maternal uncle is also severely retarded and shows the same dysmorphic facial features along with an identical fixed flexion deformity of his left index



The proband.

finger. The proband's mother has no dysmorphic features and is of normal intelligence.

### INVESTIGATIONS

The chromosome constitution in both the proband and his maternal uncle is 46,XY with no evidence of a fragile site on the X chromosome. Skull x ray performed on the proband was normal.

### Discussion

The occurrence of a similar pattern of anomalies in a

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male and his maternal uncle with normal parents and a normal karyotype makes an X linked gene defect likely. The features are distinct from the known X linked retardation syndromes.<sup>1</sup>

A search of the London Dysmorphology Database did not show a comparable case. The dysmorphic features differ from Coffin–Lowry syndrome, but show some similarities to the two brothers used as examples of Coffin–Lowry syndrome in Smith's *Recognizable patterns of human malformation*.<sup>2</sup> In retrospect, the diagnosis in these brothers is suspect.

It is possible that they are examples of the same disorder reported here, although the genital and digital defects are distinct.

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- 1 Opitz JM, Sutherland GR. Conference report. International workshop on the fragile X and X-linked mental retardation. *Am J Med Genet* 1984;17:5–94.
- 2 Smith DW. *Recognizable patterns of human malformation*. 4th ed. London: Saunders, 1988.