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Unusual Clinical Findings in Prader-Willi Syndrome Patients with Uniparental Disomy

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Eight patients with Prader-Willi syndrome (PWS), 3 males and 5 females, who were shown to have uniparental maternal disomy after investigation with polymorphic DNA marker spanning chromosome 15, were investigated clinically.

Four patients displayed in addition to the clinical signs, some unusual clinical feature: two patients showed a hairshaft anomaly, one of whom, a female, suggested a Menkes'-like condition-an investigation of copper uptake showed slightly increased values. Further investigations are in progress. One patient died unexpectedly at the age of 1 year 4 months and adenovirus was isolated postmortem in the airway secretion. Two patients presented a seizure disorder.

The mean birth weight was 2,122 (1,195-3,170) g and the gestational age was 37.1 (32-42) weeks for this group, lower than expected for PWS. Mean maternal age was elevated [31.9 (25-39) year].

Since the recent report [1] of a case of Bloom syndrome in a PWS patient led to the mapping of the Bloom syndrome gene to chromosome 15, it has been suggested that isodisomy could lead to homozygosity for recessive genes. This opens up the possibility of mapping genes in a new way. We suggest that uniparental disomy PWS patients should be investigated for unusual features to identify other monogenic disorders with a locus on chromosome 15. Isodisomy defined by multiple DNA markers indicates potential regions of candidate genes.

REFERENCES

1. Woodage et al.: Am J Hum Genet 1994; 55: 74-80.

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