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Angelman syndrome can result from uniparental paternal isodisomy

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Angelman syndrome is a rare cause of severe mental retardation with a recognisable clinical phenotype including profound speech delay, ataxic gait and behavioural disorders.

We have demonstrated a deletion of band 15q12 in approximately 50% of cases either by cytogentic of molecular genetic analysis. We have found no deletion in any of the five cases where sibs are affected in a family. In 22/22 cases the deletion occurs on the chromosome 15 inherited from the mother. In addition we have found a case which has arisen through the inheritance of two copies of one of the father's chromosomes and no inheritance of a chromosome 15 from the mother (uniparental paternal isodisomy). This was demonstrated using two multi-allelic probes CMW-1 (D15 S 24) and cMS620 (D15 S 86) which together span most of the long arm of chromosome 15.

These results mirror those found in Prader Willi syndrome in which a so far indistinguishable deletion arises on the chromosome 15 from father and in which an example of uniparental maternal heterodisomy has been found.