

## Uniparental Disomy in Beckwith-Wiedemann Syndrome

Henry et al have demonstrated that 3 of 8 cases of Beckwith-Wiedemann syndrome (BWS), a fetal overgrowth syndrome, are associated with demonstrable paternal uniparental disomy of chromosome 11. Uniparental disomy is a phenomenon in which both copies of a chromosome have been inherited from a single parent, in this case the father, with a concomitant deficiency of the maternal copy. This result was in contrast with 0 of 18 unrelated controls carrying uniparental disomy for chromosome 11. Furthermore, these authors found an overall increase in the frequency of homozygosity for several 11p15.5 markers in 21 cases of sporadic BWS, suggesting that uniparental disomy probably accounts for an even higher proportion of sporadic BWS cases than the 3 of 8 cases of unequivocal paternal uniparental disomy.

Henry I, et al. *Nature* 1991;351:665.

**Editor's comment:** Both BWS and uniparental disomy are of particular interest in the field of genomic imprinting. Many regions of the human genome, including the region of chromosome 11 implicated in BWS, appear to be imprinted, ie, they are differentially expressed depending upon whether they were inherited from the mother or the father. Because of genomic imprinting, 2 copies of an imprinted gene from a single parent, ie, uniparental disomy, might result in a strikingly different phenotype than 1 copy from each parent. The results of Henry et al support this theory and demonstrate that uniparental disomy can be associated with BWS, a cancer-predisposing genetic syndrome.

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