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Methylation and imprinting

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A third of human patients with Beckwith-Wiedemann syndrome (BWS) have lost maternal-specific methylation of the *KvDMR1* (differential methylated region) locus, a putative imprinting control region found within the *KCNQ1* gene. In an Advanced Online Publication in Nature Genetics, Fitzpatrick *et al.* provide clear evidence linking *KvDMR1* to imprinted gene expression (*Nature Genetics,* 9 September 2002, DOI:10.1038/ng988). They generated mice in which *KvDMR1* was deleted by gene-targeted homologous recombination. Paternal transmission of the deletion resulted in deregulated imprinting of the genomic locus and reactivation of genes both distal and proximal to *KvDMR1*. Paternal inheritance was also associated with reduced weight. Fitzpatrick *et al.* speculate that elevated levels of maternally expressed genes (such as *Cdkn1c*, encoding a cyclin-dependent kinase inhibitor) account for the growth defect.

References

1. Epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome.

2. *Nature Genetics*, [http://www.nature.com/ng/]