Preferential reciprocal transfer of paternal/maternal DLK1 alleles to obese children: first evidence of polar overdominance in humans.

Abstract:
DLK1 is part of the Notch signalling pathway that controls various developmental processes. A functional role for DLK1 in adipogenesis is suggested by several animal models. Interestingly, the DLK1 gene is imprinted in eutherian mammals. To study whether variations in DLK1 affect body weight in humans, we analysed 32 polymorphisms in a 109 kb genomic region encompassing DLK1 on human chromosome 14. In a study sample of 1025 French and German trio families comprised of both parents and extremely obese offspring we found a single nucleotide polymorphism (rs1802710) associated with child and adolescent obesity. Analysis of the allelic transmission pattern indicated the existence of polar overdominance, an unusual mode of non-Mendelian inheritance in humans previously known from the callipyge mutation in sheep.

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