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Modifying DiGeorge

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Patients with *del22q11* syndrome, which includes DiGeorge and velocardiofacial syndromes, present with a range of abnormalities including cardiovascular defects, thymic and parathyroid hypoplasia, facial anomalies and mental retardation. Although most patients have a common 3 Mb deletion within chromosome 22q11.2, their clinical symptoms are highly variable. In the September 25 Proceedings of the National Academy of Sciences, Ilaria Taddei and colleagues at the Baylor College of Medicine provide evidence for genetic modifiers that influence the phenotypic variability of *del22q11* syndrome (*Proc Natl Acad Sci USA* 2001, **98:**11428-11431). They studied a mouse model of the disease which harbours a deletion, *Df1*, in the corresponding region of the mouse genome. They derived *Df1* lines on different genetic backgrounds - either a pure 129SvEv background (the genetic background of the embryonic stem cell line used to generate the mice) or on a C57BL/6 background (back-crossed for nine generations). Taddei *et al.* then examined the phenotypes of *Df1*/+ embryos on the two backgrounds. They observed a lower penetrance of cardiovascular defects on the 129SvEv background (16.1%) than on the C57BL/6 background (50%). Analysis of F1 hybrid embryos indicated that allelic variation within the haploid segment does not account for the high penetrance of defects on the C57BL/6 background. Thymic anomalies were also more frequent in the congenic C57BL/6 background (42.5%) than in the inbred 129SvEv background (11.3%), but occurred independently of cardiovascular defects.

References

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