PublisherInfo		
PublisherName	:	BioMed Central
PublisherLocation	:	London
PublisherImprintName	:	BioMed Central

Gene for DiGeorge syndrome

ArticleInfo		
ArticleID	:	3997
ArticleDOI	:	10.1186/gb-spotlight-20010228-04
ArticleCitationID	:	spotlight-20010228-04
ArticleSequenceNumber	:	68
ArticleCategory	:	Research news
ArticleFirstPage	:	1
ArticleLastPage	:	2
ArticleHistory	:	RegistrationDate : 2001–02–28 OnlineDate : 2001–02–28
ArticleCopyright	:	BioMed Central Ltd2001
ArticleGrants	:	
ArticleContext	:	130592211

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DiGeorge syndrome (DGS; also known as velo-cardio-facial syndrome is associated with hemizygous deletion of a region of human chromosome 22q11, causing a range of abnormalities including cardiovascular defects, hypoplasia of the thymus and parathyroid gland, and craniofacial abnormalities. Three research groups have identified the *TBX1* gene, a member of the T-box family of transcription factors, as a key determinant of the DGS phenotype. The three reports are published in Cell, Nature Genetics and Nature. Merscher *et al*. (*Cell* 2001, **104:**619-629) and Lindsay *et al*. (*Nature* 2001, **410:**97-101) used chromosomal engineering induced using the Cre recombinase and artificial chromosome transgenesis to localize the haplosufficiency region on the mouse chromosome, chromosome 16, that corresponds to the human disease region. This region contains the *TBX1* gene, expression of which in the pharyngeal arches makes it a strong candidate gene for DGS. Both groups, together with Jerome and Papaioannou (*Nature Genetics* 2001, **27:**286-291), show that *TBX1* haploinsufficiency in mice causes cardiovascular defects and anomalies of the heart outflow tract that resemble the human syndrome. Furthermore, *TBX1-/-* mice also display thymus and parathyroid abnormalities, as seen in DGS patients. *TBX1* is the first dosage-sensitive gene to be identified in the deleted 22q11 region. It remains to be established whether *TBX1* mutation alone accounts for DiGeorge syndrome, and whether human patients differ from the mouse models in their sensitivity to *TBX1*haploinsufficiency.

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