

Title: 22q11.2 Deletion Syndrome (22q11.2DS) *GeneReview* Table 4

Authors: McDonald-McGinn DM, Dickinson KA, Emanuel BS, Zackai EH

Date: February 2013

Note: The following information is provided by the author(s) listed above and has not been reviewed by *GeneReviews* staff.

**Table 4. Genes within the DiGeorge Chromosomal Region (Centromere to Telomere)**

Gene	Function/Homology	Expression	Reference
DGCR6	Similarity to lamin g-chain & Drosophila gonadal protein	Widely expressed	Demczuk et al [1996]
LAN/DGCR2/IDD	Similarity to LDL-receptor & C-type lectin	Widely expressed	Budarf et al [1995a], Wadey et al [1995], Demczuk et al [1995]
TSK2 (serine/threonine kinase)	Serine/threonine kinase	Testis	Gong et al [1996], Goldmuntz et al [1997]
DGSI/ES2	Similarity to protein of unknown function C.elegans	Widely expressed	Gong et al [1996], Gong et al [1997], Rizzu et al [1996]
GSCL (goosecoid-like)	Homeobox protein of paired-like class	Testis; brain	Gottlieb et al [1997]
CTP (citrate transport protein)	Mitochondrial inner membrane-electroneutral exchange	Widely expressed	Heisterkamp et al [1995], Goldmuntz et al [1996]
CLTCL (clathrin heavy chain-like)	Similarity clathrin-heavy chain	Most abundant sk. muscle	Gong et al [1996], Kedra et al [1996], Sirotkin et al [1996], Holmes et al [1997]
HIRA	Homology to yeast HIR1 and HIR2	Widely expressed	Halford et al [1993b], Lorain et al [1996]
NLVCF	Unknown	Widely expressed	Funke et al [1998]
UFD1L	Similarity to yeast ubiquitin fusion degradation 1 protein	Widely expressed	Pizzuti et al [1997]
CDC45L	Similarity to other CDC45 proteins	Widely expressed	McKie et al [1998], Saha et al [1998], Shaikh et al [1999]
TMVCF	Similarity to rat protein of unknown function	Widely expressed; highest in lung	Sirotkin et al [1997b]
hCDCrel-1 (human cdc-related)	Similarity GTP-binding proteins	Unknown	Zieger et al [1997]
GP1BB	Subunit of platelet	Platelets	Budarf et al [1995b]

(glycoprotein Ib $\beta$ )	receptor von Willebrand factor		
TBX-1	Member of T box DNA family of transcription factors	Adult sk. muscle & testis; fetal tissues	Chieffo et al [1997]
CLDN5	Member of the claudin family of integral membrane proteins	Widely expressed	Sirotkin et al [1997a]
COMT (Catechol-O-methyltransferase)	Catecholamine metabolism	Widely expressed	Grossman et al [1992]
ARVCF	Novel member of catenin subfamily	Widely expressed	Sirotkin et al [1997a]
DGCR8	Microprocessor complex subunit DGCR8	Ubiquitously expressed	Gregory RI, et al., Nature. 432:235-40, 2004
T10	Unknown	Widely expressed	Halford et al [1993a]
N41 cDNA	Unknown	Widely expressed	Emanuel et al [1993]
ZNF74	RNA binding protein	Widely expressed	Aubry et al [1993], Grondin et al [1996]
LZTR-1	Similarity to leucine zipper-like domain	Widely expressed	Kurahashi et al [1995]
KLHL22	Component of the BCR ubiquitin ligase complex	Widely expressed	Maerki et al [2009]
PCQAP	Positive cofactor 2 glutamine/Q-rich-associated protein	Widely expressed	Berti et al [2001]
SNAP29	Synaptosomal-associated protein, 29kDa	Widely expressed	Steegmaier et al [1998]
CRKL	v-crk avian sarcoma virus CT10 oncogene like	Widely expressed	Ten Hoeve et al [1993]

Last updated 3-15-2012

## References

- Aubry M, Demczuk S, Desmaze C, Aikem M, Aurias A, Julien JP, Rouleau GA (1993) Isolation of a zinc finger gene consistently deleted in DiGeorge syndrome. *Hum Mol Genet* 2:1583-7
- Berti L, Mittler G, Przemeck GK, Steizer G, Gunzler B, Amati F, Conti E, Dallapiccola B, Hrabe de Angelis M, Novelli G, Meisterernst M (2001) Isolation and characterization of a novel gene from the DiGeorge chromosomal region that encodes for a mediator subunit. *Genomics* 74:320-32
- Budarf ML, Collins J, Gong W, Roe B, Wang Z, Bailey LC, Sellinger B, Michaud D, Driscoll DA, Emanuel BS (1995a) Cloning a balanced translocation associated with DiGeorge syndrome and identification of a disrupted candidate gene. *Nat Genet* 10:269-78
- Budarf ML, Konkle BA, Ludlow LB, Michaud D, Li M, Yamashiro DJ, McDonald-McGinn D, Zackai EH, Driscoll DA (1995b) Identification of a patient with Bernard-Soulier syndrome and a deletion in the DiGeorge/velocardiofacial chromosomal region in 22q11.2. *Hum Mol Genet* 4:763-6
- Chieffo C, Garvey N, Roe B, Zhang G, Silver L, Emanuel BS, Budarf ML (1997) Isolation and characterization of a gene from the DiGeorge chromosomal region (DGCR) homologous to the mouse Tbx 1 gene. *Genomics* 43:267-77
- Demczuk S, Aledo R, Zucman J, Delattre O, Desmaze C, Dauphinot L, Jalbert P, Rouleau GA, Thomas G, Aurias A (1995) Cloning of a balanced translocation breakpoint in the DiGeorge syndrome critical region and isolation of a novel potential adhesion receptor gene in its vicinity. *Hum Mol Genet* 4:551-8
- Demczuk S, Thomas G, Aurias A (1996) Isolation of a novel gene from the DiGeorge syndrome critical region with homology to *Drosophila* *gdl* and human *LAMC1* genes. *Hum Mol Genet* 5:633-8
- Emanuel BS, Driscoll D, Goldmuntz E, Baldwin S, Biegel J, Zackai EH, McDonald-McGinn D, Sellinger B, Gorman N, Williams S, et al (1993) Molecular and phenotypic analysis of the chromosome 22 microdeletion syndromes. In: Epstein, CJ (ed) *Phenotypic Mapping of Down Syndrome and Other Aneuploid Conditions*. Wiley Liss, New York, pp 207-24
- Funke B, Puech A, Saint-Jore B, Pandita R, Skjoultschi A, Morrow B (1998) Isolation and characterization of a human gene containing a nuclear localization signal from the critical region for velo-cardio-facial syndrome on 22q11. *Genomics* 53:146-54
- Goldmuntz E, Fedon J, Roe B, Budarf ML (1997) Molecular characterization of a serine/threonine kinase in the DiGeorge minimal critical region. *Gene* 198:379-86
- Goldmuntz E, Wang Z, Roe BA, Budarf ML (1996) Cloning, genomic organization and chromosomal localization of the human mitochondrial citrate transporter protein gene to chromosome 22q11 in the DiGeorge syndrome critical region. *Genomics* 33:271-6
- Gong W, Emanuel BS, Collins J, Kim DH, Wang Z, Chen F, Zhang G, Roe B, Budarf ML (1996) A transcription map of the DiGeorge and velo-cardio-facial syndrome minimal critical region on 22q11. *Hum Mol Genet* 5:1789-800
- Gong W, Emanuel BS, Galili N, Kim DH, Roe B, Driscoll DA, Budarf ML (1997) Structural and mutational analysis of a conserved gene (DGS1) from the minimal DiGeorge syndrome critical region *Hum Mol Genet* 6:267-76
- Gottlieb S, Emanuel BS, Driscoll DA, Sellinger B, Wang Z, Roe B, Budarf ML (1997) The DiGeorge syndrome minimal critical region contains a Goosecoid-like (GSCL) homeobox gene which is expressed early in human development. *Am J Hum Genet* 60:1194-201
- Gregory RI, Yan KP, Amuthan G, Chendrimada T, Doratotaj B, Cooch N, Shiekhattar R (2004) The Microprocessor complex mediates the genesis of microRNAs. *Nature* 432(7014):235-40
- Grondin B, Bazinet M, Aubry M (1996) The KRAB zinc finger gene ZNF74 encodes an RNA-binding protein tightly associated with the nuclear matrix. *J Biol Chem* 271:15458-67

Grossman MH, Emanuel BS, Budarf ML (1992) Chromosomal mapping of the human catechol-O-methyltransferase gene to 22q11.1→q11.2. *Genomics* 12:822-5

Halford S, Wadey R, Roberts C, Daw SC, Whiting JA, O'Donnell H, Dunham I, Bentley D, Lindsay E, Baldini A, et al (1993a) Isolation of a putative transcriptional regulator from the region of 22q11 deleted in DiGeorge syndrome, Shprintzen syndrome and familial congenital heart disease. *Hum Mol Genet* 2:2099-107

Halford S, Wilson DI, Daw SCM, Roberts C, Wadey R, Kamath S, Wickremasinghe A, Burn J, Goodship J, Mattei MG, et al (1993b) Isolation of a gene expressed during early embryogenesis from the region of 22q11 commonly deleted in DiGeorge syndrome. *Hum Mol Genet* 2:1577-82

Heisterkamp N, Mulder MP, Langeveld A, ten Hoeve J, Wang Z, Roe BA, Groffen J (1995) Localization of the human mitochondrial citrate transporter protein gene to chromosome 22q11 in the DiGeorge syndrome critical region. *Genomics* 29:451-6

Holmes SE, Riaz MA, Gong W, McDermid HE, Sellinger BT, Hua A, Chen F, Wang Z, Zhang G, Roe B, Gonzalez I, McDonald-McGinn DM, Zackai E, Emanuel BS, Budarf ML (1997) Disruption of the clathrin heavy chain-like gene (CLTCL) associated with features of DGS/VCFS: a balanced (21;22)(p12;q11) translocation. *Hum Mol Genet* 6:357-67

Kedra D, Peyrard M, Fransson I, Collins JE, Dunham I, Roe BA, Dumanski JP (1996) Characterization of a second human clathrin heavy chain polypeptide gene (CLH - 22) from chromosome 22q11. *Hum Mol Genet* 5:625-31

Kurahashi H, Akagi K, Inazawa J, Ohta T, Niikawa N, Kayatani F, Sano T, Okada S, Nishisho I (1995) Isolation and characterization of a novel gene deleted in DiGeorge syndrome. *Hum Mol Genet* 4:541-9

Lorain S, Demczuk S, Lamour V, Toth S, Aurias A, Roe BA, Lipinski M (1996) Structural organization of the WD repeat protein-encoding gene HIRA in the DiGeorge syndrome critical region of human chromosome 22. *Genome Res* 6:43

Maerki S, Olma MH, Staubli T, Steigermann P, Gerlich DW, Quadroni M, Sumara I, Peter M (2009) The Cul3-KLHL21 E3 ubiquitin ligase targets aurora B to midzone microtubules in anaphase and is required for cytokinesis. *J Cell Biol* 187(6):791-800

McKie JM, Wadey RB, Sutherland HF, Taylor CL, Scambler PJ (1998) Direct selection of conserved cDNAs from the DiGeorge critical region: isolation of a novel CDC45-like gene. *Genome Research* 8:834-41

Patenaude A, Ven Murthy MR, Mirault ME (2004) Mitochondrial thioredoxin system: effects of TrxR2 overexpression on redox balance, cell growth, and apoptosis. *J Biol Chem* 279(26):273012-14

Pizzuti A, Novelli G, Ratti A, Amati F, et al (1997) UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome. *Hum Mol Genet* 6:259-65

Rizzu P, Lindsay EA, Taylor C, O'Donnell H, et al (1996) Cloning and comparative mapping of a gene from the commonly deleted region of DiGeorge and Veleocardiofacial syndromes conserved in *C. elegans* Mammalian Genome 7:639-43

Saha P, Thome KC, Yamaguchi R, Hou Z, Weremowicz S, Dutta A (1998) The human homolog of *Saccharomyces cerevisiae* CDC45. *J Biol Chem* 273:18205-9

Shaikh TH, Gottlieb S, Sellinger B, Roe BA, Oakey RJ, Emanuel BS, Budarf ML (1999) Characterization of CDC45L: a gene in the 22q11.2 deletion region expressed during murine and human development. *Mammalian Genome* 10:322-6

Sirotkin H, Morrow B, DasGupta R, Goldberg R, et al (1996) Isolation of a new clathrin heavy gene with muscle-specific expression from the region commonly deleted in veleo-cardio-facial syndrome. *Hum Mol Genet* 5:617-24

Sirotkin H, Morrow B, Saint-Jore B, Puech A, et al (1997a) Identification, Characterization, and Precise Mapping of a Human Gene Encoding a Novel Membrane-Spanning Protein from the 22q11 Region Deleted in Velo-Cardio-Facial Syndrome. *Genomics* 42:245-51

Sirotkin H, Odonnell H, Dasgupta R, Halford S, et al (1997b) Identification of a new human catenin gene family member (ARVCF) from the region deleted in velo-cardio-facial syndrome. *Genomics* 41:75-83

Steegmaier M, Yang B, Yoo JS, Huang B, Shen M, Yu S, Luo Y, Scheller RH (1998) Three novel proteins of the syntaxin/SNAP-25 family. *J Biol Chem* 273:34171-9

Ten Hoeve J, Morris C, Heisterkamp N, Groffen J (1993) Isolation and chromosomal localization of CRKL, a human crk-like gene. *Oncogene*. 8:2469-74.

Wadey R, Daw S, Taylor C, Atif U, et al (1995) Isolation of a gene encoding an integral membrane protein from the vicinity of a balanced translocation breakpoint associated with DiGeorge syndrome. *Hum Mol Genet* 4:1027-33

Zieger B, Hashimoto T, Ware J (1997) Alternative expression of platelet glycoprotein Ibb mRNA from an adjacent 5' gene with an imperfect polyadenylation signal sequence. *J Clin Invest* 99:520-5