Supplementary information S1 | RefSeq genes located in the most common 3-Mb 22q11.2 microdeletion [a]

Gene Symbol [b]	Gene Name	Function Summary	Expression in HPC or forebrain [c]	Possible Genetic Link to SCZ (Ref) [d]	Abnormal brain-related phenotypes in mouse models [e]
DGCR6	DiGeorge syndrome critical region gene 6	Shares homology with the Drosophila melanogaster gonadal protein and with the gamma-1 subunit of human laminin.	Yes	Yes (1)	
PRODH	Proline dehydrogenase (oxidase) 1	Encodes for the mitochondrial enzyme proline dehydrogenase that catalyzes the first step in the degradation of L-proline, a putative neuromodulator.	Yes	Yes (1, 2)	Altered synaptic transmission, reduced PPI, impaired fear conditioning, altered cortical dopaminergic tranmission.
DGCR5	Digeorge syndrome critical region gene 5 (non-coding)	n/a			
DGCR9	DiGeorge syndrome critical region gene 9				
DGCR1	DiGeorge syndrome critical region gene 1	n/a			
DGCR2	KIAA163 gene product	Encodes a novel putative adhesion receptor protein.	Yes	Yes (3)	
DGCR11	DiGeorge syndrome critical region gene 11				
DGCR14	DiGeorge syndrome critical region gene 14	The encoded protein may be a component of spliceosomes.	Yes	Yes (4)	
TSSK2	Testis-specific serine kinase 2	Belongs to a family of serine/threonine kinases highly expressed in testis	Yes		
GSC2	Goosecoid-like	A homeodomain-containing gene, expressed in a limited number of adult tissuesand in early development.	Yes		
SLC25A1	Solute carrier family 25 (citrate transporter mitochondrial), member 1	May be involved in the regulation of chromosomal integrity	Yes		
CLTCL1	Clathrin, heavy polypeptide-like 1	Encodes a major protein of the polyhedral coat of coated pits and vesicles.			
HIRA	Hir histone cell cycle regulation defective homolog a (s. cerevisiae)	Encodes a histone chaperone that preferentially places the variant histone H3.3 in nucleosomes. May play a role in the formation of transcriptionally silent heterochomatin.	Yes		*Embryonic lethal; abnormal morphology of head, forebrain, neural tube, embryonic neuroepithelium, neural plate and altered hindbrain development.*
MRPL4	Nuclear localization signal deleted in velocardiofacial syndrome	Encodes for a mammalian mitochondrial ribosomal protein; controls in protein synthesis within the mitochondrion.	Yes		
C22orf39	Hypothetical protein loc128977	n/a			
UFD1L	Ubiquitin fusion degradation 1 like (yeast)	Encodes a protein necessary for the degradation of ubiquitinated proteins; controls the disassembly of the mitotic spindle and the formation of a closed nuclear envelope after mitosis.	Yes	Yes (5)	
CDC45L	cdc45 cell division cycle 45-like (s. cerevisiae)	Highly homologous to the Saccharomyces cerevisiae Cdc45, a protein required to the initiation of DNA replication.	Yes	Yes (6)	
CLDN5	Claudin 5	Encodes a a major cell adhesion molecule of tight junctions in brain endothelial cells.	Yes	*Yes (6)*	*Neonatal lethality; blood-brain barrier against small molecules (<8 D) was selectively affected.*
LOC15185		n/a			
SEPT5	Septin 5	Member of the septin gene family , which regulates cytoskeletal organization.	Yes		Abnormal anxiety response, reward learning and social/conspecific interaction, increased startle reflex, and prepulse inhibition, normal neuronal development and transmission.
GP1BB	Glycoprotein ib (platelet), beta polypeptide	A heterodimeric transmembrane protein that constitutes the receptor for von Willebrand factor, and mediates platelet adhesion.	Yes		
TBX1	T-box 1	Member of a family of transcription factoers that share a common DNA-binding domain, the T-box.	Yes		Decreased prepulse inhibition.
GNB1L	Guanine nucleotide binding protein (G protein), beta polypeptide 1-like	Encodes a G-protein beta-subunit-like polypeptide with WD repeats.	Yes	Yes (7)	Decreased prepulse inhibition.
C22orf29	Hypothetical protein flj21125	n/a			
TXNRD2	Thioredoxin reductase 2	A member of a family of pyridine nucleotide-disulfide oxidoreductases and a key enzyme in the regulation of the intracellular redox environment.	Yes		
COMT	Catechol-o-methyltransferase	Catalyzes the transfer of a methyl group from S-adenosylmethionine to catecholamines. One of the major degradative pathways of the catecholamine transmitters.	Yes	Yes (8–10)	Reduced clearance of cortical dopamine, increased aggression towards males, altered anxiety-related response, cognitive deficits.
ARVCF	Armadillo repeat gene deleted in velocardiofacial syndrome	A member of the catenin family, encodes a protein containing a coiled coil domain and an armadillo repeat sequence as well as a predicted nuclear-targeting sequence.	Yes	Yes (11)	
C22orf25	Chromosome 22 open reading frame 25	n/a	Yes		
DGCR8	DiGeorge syndrome critical region gene 8	Encodes for a component of the "microprocessor", a nuclear complex involved in microRNA processing.	Yes		Embryonic lethality before somite formation, abnormal dendrite morphology, abnormal spatial working memory, decreased prepulse inhibition.
TRMT2A			Yes		
RANBP1	Ran binding protein 1	Encodes a protein that binds to RAN complexed with GTP but not GDP and markedly increase GTP hydrolysis by the RanGTPase-activating protein (RanGAP1).	Yes		
ZDHHC8	Zinc finger, dhhc-type containing 8	Member of the DHHC family of palmitoyl-transferases	Yes	Yes (12, 13)	Decreased exploration in new environment, reduced prepulse inhibition, abnormal dendrite and spine morphology.
LOC15197		n/a			
RTN4R	Reticulon 4 receptor	Encodes the receptor for reticulon 4. It mediates axonal growth inhibition; may play a role in axonal plasticity in the adult CNS.	Yes	Yes (12)	Hypoactivity, impaired coordination, decreased exploration in new environment, abnormal central nervous system regeneration, axon outgrowth and plasticity.
DGCR6L	DiGeorge syndrome critical region gene 6-like	One of two genes encoding nearly identical proteins with similar expression patterns.			
PI4KAP1		n/a			
RIMBP3	Dkfzp434h735 protein	n/a			
ZNF74	Zinc finger protein 74 (cos52)	Encodes a putative transcription factor with 12 zinc finger motifs of the Kruppel/TFIIIA type.		Yes (14)	
SCARF2	Scavenger receptor class f, member 2	Similar to SCARF1/SREC-I, a scavenger receptor protein that mediates the binding and degradation of acetylated low density lipoprotein (Ac-LDL).			
KLHL22	Kelch-like 22 (drosophila)				
MED15	Trinucleotide repeat containing 7	May function as a transcriptional coactivator in RNA polymerase II transcription.	Yes		
POM121L4P	Pom121 membrane glycoprotein-like 4 pseudogene (rat)	n/a			
TMEM191A	Hypothetical protein dkfzp434n35	n/a			
PI4KA	Phosphatidylinositol 4-kinase, catalytic, alpha polypeptide	Encodes a phosphatidylinositol (PI) 4-kinase which catalyzes the first committed step in the biosynthesis of phosphatidylinositol 4,5-bisphosphate.		Yes (15)	
SERPIND1	Serpin peptidase inhibitor, clade d (heparin cofactor), member 1	Encodes serine proteinase inhibitor.			
SNAP29	Synaptosomal-associated protein, 29kda	A member of the SNAP25 gene family, encodes a protein involved in multiple membrane trafficking steps.		Yes (16)	
CRKL	v-crk sarcoma virus ct1 oncogene homolog (avian)-like	Encodes a protein kinase containing SH2 and SH3 (src homology). May be involved in the Reelin pathway.	Yes		*Double Crk and Crkl conditional KOs show impaired cerebellar foliation, Purkinje cell migration, dendritic development, and disruption of cortical layer formation.*
AIFM3	Apoptosis-inducing factor like				
LZTR1	Leucine-zipper transcription regulator 1	Encodes a member of the BTB-kelch superfamily, localized exclusively to the Golgi network.	Yes		
THAP7	THAP domain containing 7				
FLJ39582	Hypothetical protein flj39582				
MGC1673		n/a			
P2RX6	Purinergic receptor P2X, ligand-gated ion channel, 6	The protein encoded by this gene belongs to the family of P2X receptors, ATP-gated ion channels.			
SLC7A4	Solute carrier family 7 (cationic amino acid transporter), member 4				
P2RX6P		n/a			
LOC4891	Similar to chromosome 14 open reading frame 166b	n/a			
POM121L8P		n/a			
RIMBP3C	RIMS binding protein 3C	n/a			
RIMBP3B	RIMS binding protein 3B				

a. Patients with typical 3-Mb deletions that are indistinguishable by FISH analysis may exhibit differences of up to 200 Kb on each side of the breakpoint by higher resolution approaches. Since these differences can affect one or more known and predicted genes at each end, the exact complement of deleted genes may be different in each carrier.
b. The first 35 RefSeq genes listed (*DGCR6-DGCR6L*) map within the 1.5-Mb region of the deletion.
c. Experimental evidence was determined according to http://www.informatics.jax.org/expression.shtml
d. The original reference is only cited; detailed account of follow-up studies can be found at www.schizophreniagene.org
e. Italic text indicates phenotypes derived from homozygous knockout mice only.

References:
1. Liu, H. *et al.* Genetic variation at the 22q11 PRODH2/DGCR6 locus presents an unusual pattern and increases susceptibility to schizophrenia. *Proc. Natl Acad. Sci. USA* **99,** 3717–3722 (2002).
2. Jacquet, H. *et al.* PRODH mutations and hyperprolinemia in a subset of schizophrenic patients. *Hum. Mol. Genet.* **11,** 2243–2249 (2002).
3. Shifman, S. *et al.* A complete genetic association scan of the 22q11 deletion region and functional evidence reveal an association between DGCR2 and schizophrenia. *Hum. Genet.* **120**, 160–170 (2006).
4. Wang, H. *et al.* Transmission disequilibrium test provides evidence of association between promoter polymorphisms in 22q11 gene DGCR14 and schizophrenia. *J. Neural Transm.* **113**, 1551–1561 (2006).
5. De Luca, A. *et al.* Association study of a promoter polymorphism of UFD1L gene with schizophrenia. *Am. J. Med. Genet.* **105**, 529–533 (2001).
6. Sun, Z. Y., *et al.* The CLDN5 locus may be involved in the vulnerability to schizophrenia. *Eur. Psychiatry* **19**, 354–357 (2004).
7. Williams, N. M. *et al.* Strong evidence that GNB1L is associated with schizophrenia. *Hum. Mol. Genet.* **17,** 555–566 (2008).
8. Karayiorgou, M. *et al.* Genotype determining low catechol-O-methyltransferase activity as a risk factor for obsessive-compulsive disorder. *Proc. Natl Acad. Sci. USA* **94,** 4572–4575 (1997).
9. Li, T., *et al.* Preferential transmission of the high activity allele of COMT in schizophrenia. *Psychiatr. Genet.* **6**, 131–133 (1996).
10. Egan, M. F., *et al.* Effect of COMT Val108/158 Met genotype on frontal lobe function and risk for schizophrenia. *Proc. Natl. Acad. Sci. USA* **98**, 6917–6922 (2001).
11. Sanders, A.R., *et al.* Haplotypic association spanning the 22q11.21 genes COMT and ARVCF with schizophrenia. *Mol. Psychiatry* **10**, 353–365 (2005).
12. Liu, H. *et al.* Genetic variation in the 22q11 locus and susceptibility to schizophrenia. *Proc. Natl Acad. Sci. USA* **26,** 16859–16864 (2002).
13. Mukai, J. *et al.* Evidence that the gene encoding ZDHHC8 contributes to the risk of schizophrenia. *Nature Genet.* **36,** 725–731 (2004).
14. Takase, K., *et al.* Association of ZNF74 gene genotypes with age-at-onset of schizophrenia. *Schizophr. Res.* **52**, 161–165 (2001).
15. Vorstman, J. A., *et al.* Association of the PIK4CA schizophrenia-susceptibility gene in adults with the 22q11.2 deletion syndrome. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* **150B**, 430–433 (2009).
16. Saito, T., *et al.* Polymorphism in SNAP29 gene promoter region associated with schizophrenia. *Mol. Psychiatry* **6**, 193–201 (2001). Erratum in: *Mol. Psychiatry* **6**, 605 (2001).