

**Additional File 1** Genes within region of 1p36.21-1p36.33 deletion and references to gene functions

Supplementary to: N. Shiba et al., "Neuropathology of brain and spinal malformations in a case of monosomy 1p36," Acta Neuropathol Comm 2013. Correspondence: R. Hevner, Seattle Children's Research Inst., rhevner@uw.edu

Gene Symbol	Locus	Site	Type	Name	Mouse ortholog	Human Diseases	mouse phenotype	Other	Ref.
WASH5P	1p36.33	0000815..0019919*	pseudogene	WAS protein family homolog 5 pseudogene	**				
FAM138A	1p36.33	0023390..0025938*	miscRNA	family with sequence similarity 138, member A	**				
OR4F5	1p36.33	0058954..0059871*	protein coding	olfactory receptor, family 4, subfamily F, member 5	**				
OR4F16	1p36.33	0610959..0611897*	protein coding	olfactory receptor, family 4, subfamily F, member 16	**				
NCRNA00115	1p36.33	0751449..0752775*	miscRNA	non-protein coding RNA 115	**				
LOC643837	1p36.33	0766574..0782483*	miscRNA	hypothetical LOC643837	**				
FAM41C	1p36.33	0793321..0795438*	miscRNA	family with sequence similarity 41, member C	**				
FLJ39609	1p36.33	0842818..0844680*	miscRNA	similar to hCG1995469	**				
SAMD11	1p36.33	0850984..0869824*	protein coding	sterile alpha motif domain containing 11	Samd11				
NOC2L	1p36.33	0869446..0884542*	protein coding	nucleolar complex associated 2 homolog (S. cerevisiae)	Noc2l			HDAC-independent INHAT	
KLHL17	1p36.33	0885830..0890958*	protein coding	kelch-like 17 (Drosophila)	Klhl17			postsynaptic membrane	[1]
PLEKHN1	1p36.33	0891740..0900351*	protein coding	pleckstrin homology domain containing, family N member 1	Plekhn1				
HES4	1p36.33	0924207..0925333*	protein coding	hairy and enhancer of split 4 (Drosophila)	**			HES gene family	[2, 3]
ISG15 (G1P2)	1p36.33	0938742..0939782*	protein coding	ISG15 ubiquitin-like modifier	Isg15(G1p2)		viable and fertile	immune response to Mycobacteria	[4]
AGRN	1p36.33	0945366..0981355*	protein coding	agnin	Agrrn	CMS (AR)	reduced NMJ		[5, 6]
C1orf159	1p36.33	1007068..1041332*	protein coding	chromosome 1 open reading frame 159	**				
TTL10	1p36.33	1104940..1111104*	protein coding	tubulin tyrosine ligase-like family, member 10	Ttl10			glycine ligase	
TNFRSF18 (GITR)	1p36.33	1128751..1131952*	protein coding	tumor necrosis factor receptor superfamily, member 18	Tnfrsf18		immune defects	immunological self-tolerance	[7, 8]
TNFRSF4 (OX40)	1p36.33	1136569..1139375*	protein coding	tumor necrosis factor receptor superfamily, member 4	Tnfrsf4		immune defects	suppresses apoptosis	[9, 10]
SDF4	1p36.33	1142151..1157274*	protein coding	stromal cell derived factor 4	Sdf4				
B3GALT6	1p36.33	1157492..1160284*	protein coding	UDP-Gal:betaGal beta 1,3-galactosyltransferase polypeptide I	B3gal6			GAG Biosynthesis	[11]
FAM132A	1p36.33	1167696..1171965*	protein coding	family with sequence similarity 132, member A	Fam132a				
UBE2J2	1p36.33	1179155..1199097*	protein coding	ubiquitin-conjugating enzyme E2, J2 (UBC6 homolog, yeast)	Ube2j2				
LOC100128842	1p36.33	1184557..1186958*	miscRNA	hypothetical protein LOC100128842	**				
SCNN1D (ENaCd)	1p36.33	1207439..1217272*	protein coding	sodium channel, nonvoltage-gated 1, delta	**			highly expressed in pyramidal neurons	[12, 13]
ACAP3	1p36.33	1218807..1228503*	protein coding	ArfGAP with coiled-coil, ankyrin repeat and PH domains 3	Acap3				
PUSL1	1p36.33	1233857..1236920*	protein coding	polynucleotide phosphorylase 1	Pusl1				
CP5F3L	1p36.33	1236828..1249909*	protein coding	cleavage and polyadenylation specific factor 3-like	Cpsf3l			subunit of Integrator snRNA processing	[14]
GLTPD1	1p36.33	1250006..1254139*	protein coding	glycolipid transfer protein domain containing 1	Gltpd1				
TAS1R3	1p36.33	1256589..1259707*	protein coding	taste receptor, type 1, member 3	Tas1r3		taste perception defect	GPCR; calcium detection by taste	[15]
DVL1	1p36.33	1260521..1274358*	protein coding	dishevelled, dsh homolog 1 (Drosophila)	Dvl1	SJS (AR), CMT2A (AR)	abnormal social interactions,	abnormal sensorimotor gating	[16, 17]
MXRA8	1p36.33	1277934..1283778*	protein coding	matrix-remodelling associated 8	Mxra8			glia limitans / BBB	[18]
AURKAIP1	1p36.33	1298973..1300443*	protein coding	aurora kinase A interacting protein 1	Aurkaip1				
CCNL2	1p36.33	1310954..1324553*	protein coding	cyclin L2	Ccnl2				
LOC148413	1p36.33	1324795..1328199*	miscRNA	hypothetical LOC148413	**				
MRPL20	1p36.33	1327159..1332524*	protein coding	mitochondrial ribosomal protein L20	Mrpl20				
hCG_20426	1p36.33	1343665..1346513*	protein coding	hypothetical protein LOC441869	**				
TMEM88B	1p36.33	1351425..1353030*	protein coding	Transmembrane protein 88B	A230069A22Rik				
VWA1	1p36.33	1360772..1366009*	protein coding	von Willebrand factor A domain containing 1	Vwa1		peripheral nerve defects	ECM protein	[19]
ATAD3C	1p36.33	1387910..1395401*	protein coding	ATPase family, AAA domain containing 3C	**				
ATAD3B	1p36.33	1397027..1421445*	protein coding	ATPase family, AAA domain containing 3B	**				
ATAD3A	1p36.33	1437418..1459927*	protein coding	ATPase family, AAA domain containing 3A	Atad3a			Mitochondrial nucleoid organization	[20, 21]
C1orf70	1p36.33	1460602..1465597*	protein coding	chromosome 1 open reading frame 70	EG381582			Mitochondrial nucleoid organization	[20, 21]
SSU72	1p36.33	1466916..1500125*	protein coding	SSU72 RNA polymerase II CTD phosphatase homolog (S. cerevisiae)	Ssu72				
MB2	1p36.33	1540747..1555848*	protein coding	mindbomb homolog 2 (Drosophila)	Mib2		exencephaly	ubiquitin ligase for Notch ligands	[22]
MMP23B	1p36.33	1557423..1559893*	protein coding	matrix metalloproteinase 23B	Mmp23	cranial suture closure***			[23]
CDK11A (CDC2L2)	1p36.33	1560463..1580350*	protein coding	cell division cycle 2-like 2 (PITSLRE proteins)	Cdk11b			sister chromatid cohesion; apoptosis	[24-26]
RP11-345P4.4	1p36.33	1582802..1614103*	protein coding	similar to solute carrier family 35, member E2	A530082C11Rik				
MMP23A	1p36.33	1619121..1623110*	pseudogene	matrix metalloproteinase 23A (pseudogene)	**				
CDK11B (CDC2L1)	1p36.33	1624180..1645637*	protein coding	cell division cycle 2-like 1 (PITSLRE proteins)	Cdk11b			centriole duplication	[24-26]
SLC35E2	1p36.33	1653541..1667291*	protein coding	solute carrier family 35, member E2	**				
NADK	1p36.33	1672531..1699769*	protein coding	NAD kinase	Nadk				
GNB1	1p36.33	1706589..1812355*	protein coding	guanine nucleotide binding protein (G protein), beta polypeptide 1	Gnb1		microcephaly, NTDs	One of three beta subunits of G proteins	[27]
CALML6 (CAGLP)	1p36.33	1836126..1838593*	protein coding	calmodulin-like 6	**				
TMEM52	1p36.33	1838890..1840567*	protein coding	transmembrane protein 52	Tmem52				
C1orf222	1p36.33	1843256..1849228*	protein coding	chromosome 1 open reading frame 222	**				
KIAA1751	1p36.33	1874612..1925136*	protein coding	KIAA1751	2010015L04Rik				
GABRD	1p36.33	1940703..1952050*	protein coding	gamma-aminobutyric acid (GABA) A receptor, delta	Gabrd	epilepsy		pro-epileptic	[28, 29]
PRKCZ	1p36.33	1971769..2106694*	protein coding	protein kinase C, zeta	PrkcZ			lymphoid defects	aPKC (Ca++ independent)
C1orf86	1p36.33	2105777..2116074*	protein coding	chromosome 1 open reading frame 86	2610002J02Rik				[30]
LOC100128003	1p36.33	2110342..2114884*	miscRNA	hypothetical protein LOC100128003	**				
SKI	1p36.33	2149994..2229316*	protein coding	v-ski sarcoma viral oncogene homolog (avian)	Ski	facial clefting***		exencephaly, facial clefting, peripheral nerve myelination deficiency, PHPV	[31, 32]
MORN1	1p36.33-p36.32	2242556..2312853*	protein coding	MORN repeat containing 1	Morn1				
LOC100129534	1p36.33	2271716..2273960*	pseudo	small nuclear ribonucleoprotein polypeptide N pseudogene	**				
RER1	1p36.32	2313074..2326734*	protein coding	RER1 retention in endoplasmic reticulum 1 homolog (S. cerevisiae)	Rer1				
PEX10	1p36.32	2326101..2333870*	protein coding	peroxisomal biogenesis factor 10	Pex10	adrenoleukodystrophy (AR), Zellweger syndrome (AR)			[33, 34]
PLCH2	1p36.32	2397614..2426824*	protein coding	phospholipase C, eta 2	Plch2		neuron-specific isozyme	mental retardation	[35, 36]
PANK4	1p36.32	2429835..2447895*	protein coding	pantothenate kinase 4	Pank4				
HES5	1p36.32	2450044..2451544*	protein coding	hairy and enhancer of split 5 (Drosophila)	Hes5			premature differentiation	HES gene family
LOC115110	1p36.32	2471219..2474144*	miscRNA	hypothetical protein LOC115110	**				[2, 3]
TNFRSF14 (HVEM)	1p36.32	2479150..2486613*	protein coding	tumor necrosis factor receptor superfamily, member 14 (herpesvirus)	Tnfrsf14			immune defects	T cell regulation
C1orf93	1p36.32	2508109..2512762*	protein coding	chromosome 1 open reading frame 93	2810405K02Rik				[37, 38]
MMEL1 (NEP2)	1p36.32	2511941..2554289*	protein coding	membrane metallo-endopeptidase-like 1	Mmel1				
ACTRT2	1p36.32	2927906..2929327*	protein coding	actin-related protein T2	Actrt2				

FLJ42875	1p36.32	2966041..2974187*	miscRNA	hypothetical LOC440556	**				
PRDM16	1p36.32	2975604..3345045	protein coding	PR domain containing 16	Prdm16		heterochromatin disintegration	[39]	
ARHGEF16	1p36.32	3369982..3387537	protein coding	Rho guanine exchange factor (GEF) 16	Arhgef16		Zinc finger transcription factor		
MEGF6	1p36.32	3394366..3517919*	protein coding	multiple EGF-like-domains 6	Megf6				
TPRG1L	1p36.32	3531416..3536555	protein coding	tumor protein p63 regulated 1-like	Tprgl		presynaptic protein	[40]	
WDR8	1p36.32	3537199..3556497*	protein coding	WD repeat domain 8	Wdr8				
TF73	1p36.32	3558989..3639716	protein coding	tumor protein p73	Trp73		hippocampal dysgenesis, cortical apoptosis	[41, 42]	
KIAA0495	1p36.32	3642408..3653746*	protein coding	KIAA0495	**				
CCDC27	1p36.32	3658822..3678069	protein coding	coiled-coil domain containing 27	Ccdc27				
LOC388588	1p36.32	3679212..3682379	miscRNA	hypothetical LOC388588	**				
LRRc47	1p36.32	3686644..3702928*	protein coding	leucine rich repeat containing 47	Lrrc47				
KIAA0562	1p36.32	3721204..3763657*	protein coding	KIAA0562	BC046331				
DFFB	1p36.32	3763705..3791853	protein coding	DNA fragmentation factor, 40kDa, beta polypeptide (caspase-	Dffb		impaired thymic development apoptotic DNA degradation	[43]	
C1orf174	1p36.32	3795557..3806709*	protein coding	chromosome 1 open reading frame 174	A430005L14Rik				
LOC100133612	1p36.32	3806828..3821869	miscRNA	similar to hCG1815312, non-coding RNA	**				
LOC284661	1p36.32	4371971..4384604	miscRNA	hypothetical LOC284661	**				
AJAP1 (SHREW1)	1p36.32	4614965..4743711	protein coding	adherens junctions associated protein 1	Ajap1				
NPHP4	1p36.31	5845457..5975118*	protein coding	nephronophthisis 4	Nphp4	juvenile nephronophthisis (AR)	retinal degeneration	primary cilium	[44, 45]
KCNAB2	1p36.31	6008967..6083110	protein coding	potassium voltage-gated channel, shaker-related subfamily, b	Kcnab2		memory impairment	[46]	
CHD5	1p36.31	6084440..6162770*	protein coding	chromodomain helicase DNA binding protein 5	Chd5			brain development, tumor suppressor gene,	[47]
RPL22	1p36.31	6167667..6182266*	protein coding	ribosomal protein L22	Rpl22				
RNF207	1p36.31	6188776..6203946	protein coding	ring finger protein 207	Rnf207				
ICMT	1p36.21	6203840..6218631*	protein coding	isoprenylcysteine carboxyl methyltransferase	Icmt	embryonic lethal	targets proteins to cell membrane	[48, 49]	
HES3	1p36.31	6226849..6228225	protein coding	hairly and enhancer of split 3 (Drosophila)	Hes3	premature differentiation	HES gene family	[2, 3]	
GPR153	1p36.31	6231443..6243622*	protein coding	G protein-coupled receptor 153	Gpr153				
ACOT7 (BACH)	1p36.31	6246919..6376413*	protein coding	acyl-CoA thioesterase 7	Acot7				
HES2	1p36.31	6397883..6402566*	protein coding	hairly and enhancer of split 2 (Drosophila)	Hes2		MTLE	[50]	
ESPN	1p36.31	6407435..6443591	protein coding	espin	Espn (je)	congenital hearing loss (AD, A jerker deafness	expressed in developing nervous system	[2, 3]	
TNFRSF25 (DR3)	1p36.31	6443798..6448842*	protein coding	tumor necrosis factor receptor superfamily, member 25	Tnfrsf25	rheumatoid arthritis	actin bundling	[51, 52]	
PLEKHG5	1p36.31	6448739..6502656*	protein coding	pleckstrin homology domain containing, family G (with RhoGe	Plekhg5	immune defects	T cell apoptosis and self-tolerance	[53-55]	
NOL9	1p36.31	6507792..6537168*	protein coding	nucleolar protein 9	Nol9	lower motor neuron disease (AR)	activates NF kappa B	[56]	
TAS1R1	1p36.31	6538021..6562404	protein coding	taste receptor, type 1, member 1	Tas1r1		taste perception defect	GPCR; sweet and umami tastes	[57]
ZBTB48 (HKR3)	1p36.31	6562698..6571926	protein coding	zinc finger and BTB domain containing 48	Zbtb48				
KLHL21	1p36.31	6573371..6585516*	protein coding	kelch-like 21 (Drosophila)	Klhl21				
PHF13	1p36.31	6596332..6606643	protein coding	PHD finger protein 13	Phf13				
THAP3	1p36.31	6607839..6618232	protein coding	THAP domain containing, apoptosis associated protein 3	Thap3				
DNAJC11	1p36.31	6616818..6684460*	protein coding	DnaJ (Hsp40) homolog, subfamily C, member 11	Dnajc11				
CAMTA1	1p36.31-p36.23	6767971..7752351	protein coding	calmodulin-binding transcription activator 1	Camta1				

AD autosomal dominant, AR autosomal recessive, BBB blood brain barrier, CMS Congenital myasthenic syndrome, CMT2A Charcot-Marie-Tooth disease type 2A, ECM extracellular matrix protein, GABA Gamma-aminobutyric acid, GPCR G protein-coupled receptor, HDAC histone deacetylases, IHAT inhibitor of histone acetyltransferase, MTLE mesial temporal epilepsy, PHPV persistent hyperplastic primary vitreous, SJS Schwartz-Jampel syndrome, SLS Senior-Loken syndrome

\* complement=minus strand, \*\* not exist, \*\*\* putative causative gene

## References for Additional File 1 Table

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