



<i>e!</i>	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970	
TSEN34 <i>e!</i>	Pontocerebellar hypoplasia type 2	OrphaNet	OrphaNet:2524	
CNOT3 <i>e!</i>	Precursor T-cell acute lymphoblastic leukemia	OrphaNet	OrphaNet:99861	



### Direct effect on transcript

#### Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
AC009892.5	splice donor variant	ENST00000432400 ?		?					1
LILRB1 <i>e!</i>	missense variant	ENST00000324602	NM_001081637.1	ENSP00000315997	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000396315 ?		ENSP00000379608	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000396331	NM_006669.5	ENSP00000379622	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000427581 ?		ENSP00000395004	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000396327	NM_001081638.2	ENSP00000379618	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000421584 ?		ENSP00000410165	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000396317	NM_001278398.1	ENSP00000379610	V/L	Gtg/Ctg	?	?	1
LILRB1 <i>e!</i>	missense variant	ENST00000396332	NM_001081639.2	ENSP00000379623	V/L	Gtg/Ctg	?	?	1

### Direct effect on regulation

#### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRB1 <i>e!</i>	ENST00000462628 <i>e!</i>	ILMN_1708248 <i>e!</i>	blood	9.02×10 <sup>-31</sup> (p-value)	MuTHER consortium 	1
LILRB1 <i>e!</i>	ENST00000396315 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000427581 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000396327 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000396317 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000480375 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000324602 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000421584 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000396332 <i>e!</i>					
LILRB1 <i>e!</i>	ENST00000396331 <i>e!</i>					
LILRB1 <i>e!</i>	?	ENSG00000104972 <i>e!</i>	testis	3.99×10 <sup>-6</sup> (p-value)	GTE Portal V6 	1

### Putative effect on regulation

#### FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000344796 <i>e!</i>	1	ENST00000418536 <i>e!</i> , ENST00000396321 <i>e!</i>	LILRB1 <i>e!</i>

#### ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024800 <i>e!</i>		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233565 <i>e!</i>	2	ENCP00000024749	AC012314.20 <i>e!</i> AC012314.20 <i>e!</i>
		ENCP00000024780	TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i>
		ENCP00000024781	TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i>
		ENCP00000024748	TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i> TSEN34 <i>e!</i>
		ENCP00000024778	TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i>
		ENCP00000024742	MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i>
		ENCP00000024745	MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i> MBOAT7 <i>e!</i>
		ENCP00000024733	CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i> CNOT3 <i>e!</i>
		ENCP00000024804	LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i>
		ENCP00000024777	TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i> TTYH1 <i>e!</i>
		ENCP00000024741	TMC4 <i>e!</i> TMC4 <i>e!</i> TMC4 <i>e!</i>

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		ENCP00000024728	AC012314.8 <i>e!</i>
			AC012314.8 <i>e!</i>
ENCE00000233676 <i>e!</i>	1	ENCP00000024766	AC008984.6 <i>e!</i>
			AC008984.6 <i>e!</i>
		ENCP00000024795	KIR3DX1 <i>e!</i>
			KIR3DX1 <i>e!</i>
			KIR3DX1 <i>e!</i>
			KIR3DX1 <i>e!</i>
		ENCP00000024826	NLRP2 <i>e!</i>
			NLRP2 <i>e!</i>
			NLRP2 <i>e!</i>
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			NLRP2 <i>e!</i>
		ENCP00000024805	LILRB1 <i>e!</i>
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			LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
		ENCP00000024809	AC009892.9 <i>e!</i>
		ENCP00000024796	AC009892.2 <i>e!</i>
			AC009892.2 <i>e!</i>
		ENCP00000024793	LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>
		ENCP00000024801	LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
			LILRB1 <i>e!</i>
ENCE00000233601 <i>e!</i>	1	ENCP00000024767	LILRA4 <i>e!</i>
			LILRA4 <i>e!</i>
			LILRA4 <i>e!</i>
			LILRA4 <i>e!</i>
			AC008984.2 <i>e!</i>
			AC008984.2 <i>e!</i>
		ENCP00000024744	MBOAT7 <i>e!</i>
			MBOAT7 <i>e!</i>
			MBOAT7 <i>e!</i>
			MBOAT7 <i>e!</i>
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			MBOAT7 <i>e!</i>
			MBOAT7 <i>e!</i>
			MBOAT7 <i>e!</i>
		ENCP00000024775	AC008746.10 <i>e!</i>
			AC008746.10 <i>e!</i>
		ENCP00000024779	TTYH1 <i>e!</i>
			TTYH1 <i>e!</i>
			TTYH1 <i>e!</i>
			TTYH1 <i>e!</i>
		ENCP00000024792	LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>
			LAIR2 <i>e!</i>

ENCP00000024814

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ENCP00000024815

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ENCP00000024774

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LAIR1 e!

ENCE00000233679 e!

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ENCP00000024815

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ENCP00000024796

AC009892.2 e!  
AC009892.2 e!

ENCP00000024766

AC008984.6 e!  
AC008984.6 e!

ENCP00000024793

LAIR2 e!  
LAIR2 e!  
LAIR2 e!

		ENCP00000024826	LAIR2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i> NLRP2 <i>e!</i>
		ENCP00000024801	LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i> LILRB1 <i>e!</i>
		ENCP00000024795	KIR3DX1 <i>e!</i> KIR3DX1 <i>e!</i> KIR3DX1 <i>e!</i> KIR3DX1 <i>e!</i>
ENCE00000233738 <i>e!</i>	1	ENCP00000024793	LAIR2 <i>e!</i> LAIR2 <i>e!</i> LAIR2 <i>e!</i> LAIR2 <i>e!</i>

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348881 <i>e!</i> (open chromatin region)	7	blood (GM12878) blood (K562) A549	H3K4me3, H3K27ac, H3K4me2 H3K27me3 H3K27me3
ENSR00000348884 <i>e!</i> (promoter)	1	cervix (HeLa-S3) embryonic stem cell (H1ESC) blood (GM12878) A549 blood (K562)	H3K27me3 DNase1 Yy1, PU1, PolII, BATF, DNase1, IRF4, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, H3K4me2, H3K9ac H3K27me3 H3K27me3

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC009892.10 <i>e!</i>	downstream gene variant	1067	ENST00000456337 <i>e!</i>	?	?	3
AC009892.5 <i>e!</i>	upstream gene variant, downstream gene variant	53	ENST00000432400 <i>e!</i>	?	?	32
AC245036.1 <i>e!</i>	downstream gene variant, upstream gene variant	678	ENST00000578908 <i>e!</i>	?	?	5
LILRA1 <i>e!</i>	downstream gene variant	1211	ENST00000495417 <i>e!</i>	?	?	2
LILRA1 <i>e!</i>	downstream gene variant	1447	ENST00000618665 <i>e!</i>	?	ENSP00000479482 <i>e!</i>	2
LILRA1 <i>e!</i>	downstream gene variant	2156	ENST00000453777 <i>e!</i>	NM_001278318.1	ENSP00000413715 <i>e!</i>	4
LILRA1 <i>e!</i>	downstream gene variant	1215	ENST00000477255 <i>e!</i>	?	?	2
LILRA1 <i>e!</i>	downstream gene variant	3171	ENST00000251372 <i>e!</i>	NM_006863.3	ENSP00000251372 <i>e!</i>	3
LILRA1 <i>e!</i>	downstream gene variant	2369	ENST00000473156 <i>e!</i>	?	?	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	859	ENST00000324602 <i>e!</i>	NM_001081637.1	ENSP00000315997 <i>e!</i>	3
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	315	ENST00000421584 <i>e!</i>	?	ENSP00000410165 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	859	ENST00000396331 <i>e!</i>	NM_006669.5	ENSP00000379622 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant	2360	ENST00000396321 <i>e!</i>	?	ENSP00000379614 <i>e!</i>	2
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	315	ENST00000396317 <i>e!</i>	NM_001278398.1	ENSP00000379610 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	859	ENST00000462628 <i>e!</i>	?	?	2
LILRB1 <i>e!</i>	upstream gene variant	2360	ENST00000630741 <i>e!</i>	?	ENSP00000485956 <i>e!</i>	2
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	238	ENST00000480375 <i>e!</i>	?	?	6

LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	859	ENST00000396332 <i>e!</i>	NM_001081639.2	ENSP00000379623 <i>e!</i>	3
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	859	ENST00000396327 <i>e!</i>	NM_001081638.2	ENSP00000379618 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	2230	ENST00000480257 <i>e!</i>	?	?	6
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	315	ENST00000396315 <i>e!</i>	?	ENSP00000379608 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	102	ENST00000473412 <i>e!</i>	?	?	7
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	201	ENST00000427581 <i>e!</i>	?	ENSP00000395004 <i>e!</i>	4
LILRB1 <i>e!</i>	upstream gene variant, downstream gene variant	86	ENST00000487425 <i>e!</i>	?	?	4
LILRB1 <i>e!</i>	upstream gene variant	2322	ENST00000418536 <i>e!</i>	?	ENSP00000391514 <i>e!</i>	2

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC009892.10 <i>e!</i>	ENST00000456337 <i>e!</i>	?	?	3
LILRA1 <i>e!</i>	ENST00000251372 <i>e!</i>	NM_006863.3	ENSP00000251372 <i>e!</i>	1
LILRA1 <i>e!</i>	ENST00000453777 <i>e!</i>	NM_001278318.1	ENSP00000413715 <i>e!</i>	1
LILRA1 <i>e!</i>	ENST00000473156 <i>e!</i>	?	?	1
LILRB1 <i>e!</i>	ENST00000396317 <i>e!</i>	NM_001278398.1	ENSP00000379610 <i>e!</i>	3
LILRB1 <i>e!</i>	ENST00000396321 <i>e!</i>	?	ENSP00000379614 <i>e!</i>	6
LILRB1 <i>e!</i>	ENST00000324602 <i>e!</i>	NM_001081637.1	ENSP00000315997 <i>e!</i>	4
LILRB1 <i>e!</i>	ENST00000396327 <i>e!</i>	NM_001081638.2	ENSP00000379618 <i>e!</i>	4
LILRB1 <i>e!</i>	ENST00000630741 <i>e!</i>	?	ENSP00000485956 <i>e!</i>	6
LILRB1 <i>e!</i>	ENST00000418536 <i>e!</i>	?	ENSP00000391514 <i>e!</i>	6
LILRB1 <i>e!</i>	ENST00000396331 <i>e!</i>	NM_006669.5	ENSP00000379622 <i>e!</i>	12
LILRB1 <i>e!</i>	ENST00000421584 <i>e!</i>	?	ENSP00000410165 <i>e!</i>	3
LILRB1 <i>e!</i>	ENST00000396315 <i>e!</i>	?	ENSP00000379608 <i>e!</i>	3
LILRB1 <i>e!</i>	ENST00000396332 <i>e!</i>	NM_001081639.2	ENSP00000379623 <i>e!</i>	4
LILRB1 <i>e!</i>	ENST00000462628 <i>e!</i>	?	?	5
LILRB1 <i>e!</i>	ENST00000427581 <i>e!</i>	?	ENSP00000395004 <i>e!</i>	3

#### 3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRA1 <i>e!</i>	ENST00000618665 <i>e!</i>	?	ENSP00000479482 <i>e!</i>	1
LILRA1 <i>e!</i>	ENST00000251372 <i>e!</i>	NM_006863.3	ENSP00000251372 <i>e!</i>	2
LILRA1 <i>e!</i>	ENST00000453777 <i>e!</i>	NM_001278318.1	ENSP00000413715 <i>e!</i>	2
LILRB1 <i>e!</i>	ENST00000396317 <i>e!</i>	NM_001278398.1	ENSP00000379610 <i>e!</i>	1
LILRB1 <i>e!</i>	ENST00000324602 <i>e!</i>	NM_001081637.1	ENSP00000315997 <i>e!</i>	2
LILRB1 <i>e!</i>	ENST00000396327 <i>e!</i>	NM_001081638.2	ENSP00000379618 <i>e!</i>	2
LILRB1 <i>e!</i>	ENST00000396331 <i>e!</i>	NM_006669.5	ENSP00000379622 <i>e!</i>	2
LILRB1 <i>e!</i>	ENST00000421584 <i>e!</i>	?	ENSP00000410165 <i>e!</i>	1
LILRB1 <i>e!</i>	ENST00000396315 <i>e!</i>	?	ENSP00000379608 <i>e!</i>	1
LILRB1 <i>e!</i>	ENST00000396332 <i>e!</i>	NM_001081639.2	ENSP00000379623 <i>e!</i>	2
LILRB1 <i>e!</i>	ENST00000427581 <i>e!</i>	?	ENSP00000395004 <i>e!</i>	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC009892.5 <i>e!</i>	ENST00000432400 <i>e!</i>	?	1
LILRA1 <i>e!</i>	ENST00000477255 <i>e!</i>	?	1
LILRA1 <i>e!</i>	ENST00000495417 <i>e!</i>	?	1
LILRA1 <i>e!</i>	ENST00000473156 <i>e!</i>	?	2
LILRB1 <i>e!</i>	ENST00000462628 <i>e!</i>	?	4
LILRB1 <i>e!</i>	ENST00000487425 <i>e!</i>	?	2



