eTable 1 Information of *TRIP12* mutations have been identified

Position	Nucleotide change †	Amino-acid change	Mutation Type	Domain	Reference
chr2:230744782	c.14C>T	p.(Pro5Leu)	missense	IDR	LOVD
chr2:230724181	c.208C>T	p.(Arg70Ter)	nonsense	IDR	HGMD
chr2:230724115-230724116	c.273dup	p.(Pro92ThrfsTer8)	frameshift	IDR	ClinVar
chr2:230724029	c.360del	p.(Gln120HisfsTer3)	frameshift	IDR	ClinVar
chr2:230723928-230723929	c.460_461del	p.(Ser154PhefsTer10)	frameshift	IDR	ClinVar
chr2:230701696	c.1012C>T	p.(Arg338Ter)	nonsense	IDR	LOVD
chr2:230701654	c.1054C>T	p.(Arg352Ter)	nonsense	IDR	LOVD
chr2:230695547	c.1145-10del	-	Frameshift *	IDR	ClinVar
chr2:230695539	c.1145-2A>C	-	splicing	IDR	HGMD
chr2:230695490	c.1192G>T	p.(Glu398Ter)	nonsense	IDR	ClinVar
chr2:230693992-230693995	c.1220_1223del	p.(Ala408GlyfsTer77)	frameshift	IDR	LOVD
chr2:230693985-230693986	c.1229dup	p.(Leu411SerfsTer19)	frameshift	IDR	ClinVar
chr2:230683078	c.1455+2T>C	-	splicing	IDR	ClinVar
chr2:230679895	c.1507C>T	p.(Arg503Ter)	nonsense	ARM	ClinVar
chr2:230679862	c.1540C>T	p.(Arg514Ter)	nonsense	ARM	ClinVar
chr2:230678967-230678991	c.1638_1662del	p.(Glu547LysfsTer16)	frameshift	ARM	ClinVar
chr2:230678715	c.1713del	p.(Glu571AspfsTer5)	frameshift	ARM	ClinVar
chr2:230675752	c.1921C>T	p.(Gln641Ter)	nonsense	ARM	LOVD
chr2:230675713	c.1960C>T	p.(Gln654Ter)	nonsense	ARM	LOVD

chr2:230675698-230675699	c.1974_1975insT	p.(Thr659TyrfsTer57)	frameshift	ARM	ClinVar
chr2:230672494	c.2282C>T	p.(Ala761Val)	missense	WWE	LOVD
chr2:230670506-230670510	c.2361_2365dup	p.(Asp791ValfsTer17)	frameshift	WWE	LOVD
chr2:230670507	c.2365del	p.(Asp789ThrfsTer15)	frameshift	WWE	LOVD
chr2:230670443	c.2425+4dup	-	splicing	WWE	LOVD
chr2:230668791	c.2578A>T	p.(Arg860Ter)	nonsense	-	LOVD
chr2:230668284	c.2773+3A>G	-	splicing	-	LOVD
chr2:230667008	c.2941C>T	p.(Gln981Ter)	nonsense	-	ClinVar
chr2:230666969-230666970	c.2979dup	p.(Gly994ArgfsTer5)	frameshift	-	ClinVar
chr2:230666967	c.2981+1delG	-	splicing	-	HGMD
chr2:230664098	c.2983C>T	p.(Arg995Ter)	nonsense	-	ClinVar
chr2:230663590	c.3257+1G>A	-	splicing	-	ClinVar
chr2:230662539-230662540	c.3265dup	p.(Ile1089AsnfsTer2)	frameshift	-	ClinVar
chr2:230662530	c.3275G>A	p.(Trp1092Ter)	nonsense	-	ClinVar
chr2:230662444	c.3361C>T	p.(Gln1121Ter)	nonsense	-	ClinVar
chr2:230661451-230661452	c.3446_3447del	p.(Ser1149Ter)	nonsense	-	ClinVar
chr2:230661366	c.3532del	p.(Asp1178MetfsTer3)	frameshift	-	ClinVar
chr2:230661344	c.3554del	p.(Arg1185AsnfsTer2)	frameshift	-	ClinVar
chr2:230661315	c.3583del	p.(Ser1195LeufsTer24)	frameshift	-	ClinVar
chr2:230659947	c.3691C>T	p.(Gln1231Ter)	nonsense	-	ClinVar
chr2:230659894	c.3743+1G>T	-	splicing	-	ClinVar

chr2:230659894	c.3743+1G>A	-	splicing	-	ClinVar
chr2:230657845-230657846	c.3759_3760del	p.(Gly1254IlefsTer36)	frameshift	-	ClinVar
chr2:230657794	c.3811C>T	p.(Gln1271Ter)	nonsense	-	ClinVar
chr2:230657776-230657777	c.3828_3829del	p.(Ala1277LysfsTer13)	frameshift	-	ClinVar
chr2:230657754	c.3851del	p.(Gly1284ValfsTer20)	frameshift	-	ClinVar
chr2:230657702	c.3903C>A	p.(Tyr1301Ter)	nonsense	-	LOVD
chr2:230656688-230656691	c.4081_4084dup	p.(Val1362GlyfsTer6)	frameshift	-	LOVD
chr2:230656679	c.4093C>T	p.(Gln1365Ter)	nonsense	-	ClinVar
chr2:230656628-230656629	c.4143dup	p.(Asn1382GlnfsTer18)	frameshift	-	ClinVar
chr2:230654354-230654376	c.4421_4443del	p.(Leu1474GlnfsTer9)	frameshift	-	ClinVar
chr2:230654322	c.4470+5G>A	-	splicing	-	ClinVar
chr2:230654326	c.4470+1G>A	-	splicing	-	ClinVar
chr2:230653626	c.4501del	p.(Ser1501ValfsTer8)	frameshift	-	ClinVar
chr2:230653512	c.4613+2T>G	-	splicing	-	ClinVar
chr2:230652322	c.4669G>C	p.(Asp1557His)	missense	HECT	LOVD
chr2:230652313	c.4678C>T	p.(Arg1560Ter)	nonsense	HECT	ClinVar
chr2:230652312	c.4679G>A	p.(Arg1560Gln)	missense	HECT	ClinVar
chr2:230652230	c.4761T>A	p.(Asp1587Glu)	missense	HECT	ClinVar
chr2:230652219	c.4770+2T>C	-	splicing	HECT	ClinVar
chr2:230652221	c.4770del	p.(Lys1590AsnfsTer4)	frameshift	HECT	ClinVar
chr2:230650558	c.4784G>A	p.(Arg1595Gln)	missense	HECT	ClinVar
chr2:230643607-230643610	c.4966_4969del	p.(Thr1656LeufsTer39)	frameshift	HECT	ClinVar

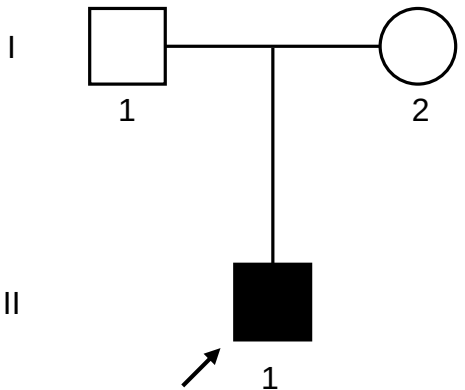
chr2:230643603-230643607	c.4966_4969del	p.(Thr1656LeufsTer39)	frameshift	HECT	ClinVar
chr2:230643298	c.4969_4973del	p.(Leu1657GlnfsTer25)	frameshift	HECT	LOVD
chr2:230642100-230642101	c.4990del	p.(Gln1664LysfsTer32)	frameshift	HECT	ClinVar
chr2:230636299	c.5234dup	p.(Ala1746SerfsTer18)	frameshift	HECT	ClinVar
chr2:230636242	c.5519C>T	p.(Ser1840Leu)	missense	HECT	LOVD
chr2:230636234	c.5576C>T	p.(Pro1859Leu)	missense	HECT	ClinVar
chr2:230634035	c.5583+1G>A	-	splicing	HECT	LOVD
chr2:230632449	c.5590C>T	p.(Gln1864Ter)	nonsense	HECT	LOVD
chr2:230632366-230632379	c.5800C>A	p.(Pro1934Thr)	missense	HECT	LOVD
chr2:230632352	c.5863_5876dup	p.(Val1960LeufsTer2)	frameshift	HECT	LOVD
chr2:230632293	c.5897C>T	p.(Pro1966Leu)	missense	HECT	ClinVar

† DNA mutation Numbering is based on cDNA sequence (GenBank no. NM_004238.3), with nucleotide +1 corresponding to the first nucleotide of the ATG translation initiation codon.
* NM_001348323.3 was used as reference in the ClinVar database, and the deletion was described as a frameshift variation.

eTable 2 Primer pairs used in Sanger sequencing and RT-qPCR

Primer name	Primer sequence(5'-3')		Purpose	Product size (bp)
	Forward	Reverse		
TRIP12-E12	ATTGCTGCCAGAGTATCACG	ACAGAGCGAGACTCCCATCT	PCR/Sequence	232
TRIP12-E20	AAAGGCATGTACGAATGGATC	CAGTTCCTAAACACAGCCCA	PCR/Sequence	343
TRIP12-mRNA	GGAGATGTTGTCACGGAG	TGAACAGCTAAAGTTGGACA	PCR/Sequence	426
GAPDH	GTGAAGGTCGGAGTCAACG	TGAGGTCAATGAAGGGGTC	RT-qPCR	112
TRIP12-q	CCTGGACCTTCTGGATTACA	GTCTTTTGGCGTGTGCTC	RT-qPCR	117
ASXL1-q	ATGCTCCAATGACACCAAAAC	AAAACAACCCCTCTCCTCCTC	RT-qPCR	138
USP7-q	CACACCAAAAAAGCGTAGGAT	TCAATACGACGACTGAACGACT	RT-qPCR	136
PTF1A-q	AGAAGGTCATCATCTGCCATC	TTCCTTGAGTTGTTTTTCATCAGT	RT-qPCR	119
SOX6-q	GAAACAACGGCAGCAAATG	ATGTGACCCTGAACCTGGAT	RT-qPCR	129
RNF168-q	TGAAAAGTGATGAGGAACTGG	TCAGACTTGGGTGTAACTGGAT	RT-qPCR	121
SMARCE1-q	TCAAATAGAGGAACGACACCAG	CTACTTTCAGACCGCACAACC	RT-qPCR	98
CDKN2A-q	GAATAGTTACGGTCGGAGGC	ACGGGTCGGGTGAGAGT	RT-qPCR	123
NAE1-q	TTCCTGAACTGAGAGAACATTTTC	GGTATTCGTCCATTTGTTTCAC	RT-qPCR	138
PARP1-q	CAGGAGTCAAGAGTGAAGGAAAG	AGATCAGGTCGTTCTGAGCC	RT-qPCR	138

Family 1



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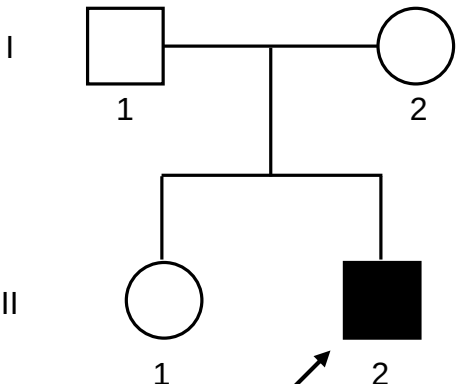
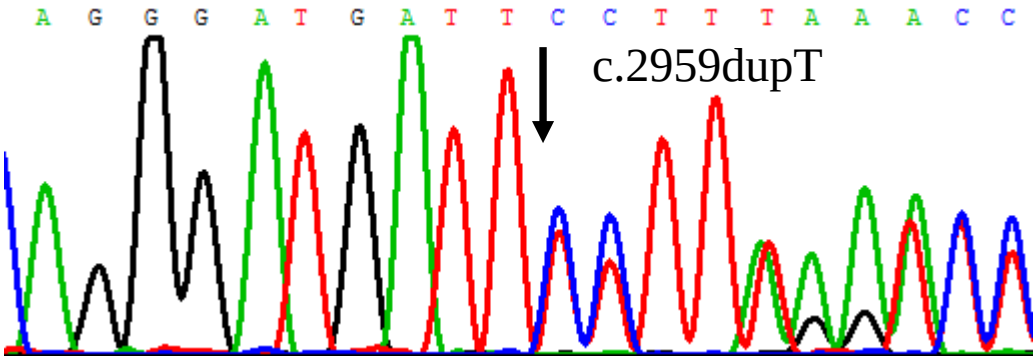


Family 1

Proband



Control

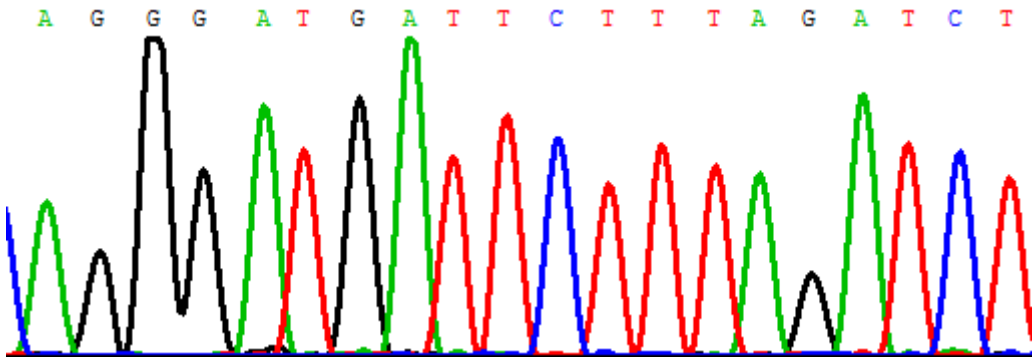


Figure legends

eFigure 1 Pedigrees of the two families. The probands indicated by the arrow in the family pedigrees.

eFigure 2 DNA sequencing results of the *TRIP12* gene in the family 1. Arrow indicate mutation site.