

Table S1. Target enrichment kits, platform and WES statistics

	<b>patient 1</b>	<b>patient 2</b>
<b>WES enrichment kit</b>	Twist Human Core Exome Kit v.1.3 (Twist Bioscience)	Twist Human Core Exome+RefSeq Panel (Twist Biosciences)
<b>Sequencing platform</b>	Nextseq 500 sequencer (Illumina)	NextSeq 550 sequencer (Illumina)
<b>Target regions covered &gt;10x</b>	99,60%	96,7%
<b>Target regions covered &gt;20x</b>	99,40%	94,5%
<b>Average depth on target</b>	108.8X	47X
<b>Alignment and Variant calling</b>	Burrows-Wheeler Aligner (BWA 0.7.17) and GATK v.4.1	BWA-mem tool and GATK v4.1.2
<b>Annotation and Prioritization based on ACMG guidelines 2015</b>	Annovar	eVAI-enGenome

Table S2. Oligonucleotides used for qRT-PCR experiments and for Actinomycin D assay

	Forward	Reverse
<b>primer pair 1</b>	GGACAAATTTAATCTTACTGGAC	CTCCTTGCTGGTACTTTTCC
<b>primer pair 2</b>	TGTGTACTGTGAGAACCAGCC	CATGTGAGGGAACCAGTGC
<b>primer pair 3</b>	CTCCCATTGGCCTTCAGAC	AAGAGCATGTGAGGGAACC
<b>primers wt 1</b>	GACAAATTTAATCTTACTGGACT	CTCCTTGCTGGTACTTTTCC
<b>primers wt 2</b>	AGACATCCCAGGTGAAGCCA	AAGAGCATGTGAGGGAACC
<b>primers <i>Leu39Arg</i></b>	GACAAATTTAATCTTACTGGACG	CTCCTTGCTGGTACTTTTCC
<b>primers <i>Met132fs</i></b>	AGACATCCCTGGTGAAGCTCT	AAGAGCATGTGAGGGAACC
<b>primers <i>CSNK2B</i></b>	GTACCAGCAAGGAGACTTTC	AAGAGCATGTGAGGGAACC
<b>primers <math>\beta</math>-Actin</b>	CACCTTCTACAATGAGCTGC	CACAGCCTGGATAGCAACG

Table S3. *CSNK2B* mutations identified in patient 1 and 2 and their predicted functional impact, based on ACMG guideline 2015, using Varsome (<https://varsome.com>) and *in silico* tools

	patient 1	patient 2
<b>Genomic position (GRCh37/hg19)</b>	chr6:31635688T>G	chr6:31637112_31637122delAGGTGAAGCCA
<b>Variant (NM_001320.7)</b>	c.116T>G	c.384_394del
<b>Predicted protein change</b>	p.Leu39Arg	pMet132LeufsTer110
<b>Variant type</b>	missense	fremeshift
<b>Zygoty</b>	heterozygous	heterozygous
<b>Inheritance</b>	<i>de novo</i>	<i>de novo</i>
<b>Predicted effect (ACMG guidelines 2015) and using Varsome</b>	Pathogenic: PM1, PM2, PP2, PP3, PS2	Pathogenic: PVS1, PS2
<b>PaPI</b>	1 (DAMAGING)	0.968 (DAMAGING)
<b>PolyPhen-2</b>	1 (DAMAGING)	NA
<b>SIFT</b>	0 (DAMAGING)	NA
<b>MutationTaster</b>	DELETERIOUS	DELETERIOUS
<b>DANN</b>	0.997 (DAMAGING)	NA