Patient		1		2	4		5
Gene		*NEB*		*NEB*	*NEB*		*ACTA1*
Locus		chr2:152424649_152424650	chr2:152473973	chr2:152394412	chr2:152424649_152424650	chr2:152547236	chr1:229567587
Exon		113	75	140	113	24	6
Sequence variants		c.17779_17780delTA (p.Tyr5927HisfsX17)	c.11086A>C (p.Thr3696Pro)	c.21076C>T (p.Arg7026Ter)	c.17779_17780delTA (p.Tyr5927HisfsX17)	c.2310+5G>A	c.871A>T (p.Ile291Phe)
State		heterozygote	heterozygote	heterozygote	heterozygote	heterozygote	heterozygote
RefSeq		NM_001271208.1	NM_001271208.1	NM_001271208.1	NM_001271208.1	NM_001271208.1	NM_001100.3
European (non-finnish) Population Frequency	**gnomAD**	NF	NF	0,01%	NF	NF	NF
	ExAC	NF	NF	NF	NF	NF	NF
Predictors *in silico*	***Mutation taster***	DC	DC	DC	DC	DC	DC
	SIFT	-	T	-	-	*	P
	Polyphen-2	-	PsD	-	-	*	PsD
References		*novel*	*novel*	Lehtokari *et al*, 2014	*novel*	*novel*	Laing *et al*,2009
ACMG evidence of pathogenity		Pathogenic	Variant of Uncertain Significance	Pathogenic	Pathogenic	Pathogenic	Pathogenic
Inheritance		Inherited from father	Inherited from mother	Inherited from father	Inherited from mother	Inherited from father	*de novo*

NF=Not found P= Pathogenic B=Benign PrD=Probably Damaging
DC= Disease causing T= Tolerated N=Neutral PsD= Possibly Damaging
*Splicing predictors corresponding to this variant are shown in Table 4.