

Supplementary table S2. Segregation of identified variants in families

Family	Gene	variant	Proband		Mother		Father		Additional family members		
			Phenotype	Genotype	Phenotype	Genotype	Phenotype	Genotype	Relative	Phenotype	Genotype
Solved families											
TB533/ TB621	MFRP	Exon 1 deletion	affected	v/v	unaffected	NT	unaffected	NT	Brother	affected	v/v
AA1	SOX2	c.368A>G; p.(Asp123Gly)	affected	v/+	unaffected	v/+	unaffected	+/+	-	-	-
NGC9	RARB	c.1196T>C; p.(Met399Thr)	affected	v/+ (de novo)	unaffected	+/+	unaffected	+/+	-	-	-
NGC10	SMO	c.1427A>G; p.(Asn476Ser)	affected	v/+	unaffected	NT	unaffected	v/+	-	-	-
Possibly solved families											
AW1	SHH	c.868G>T; p.(Gly290Cys)	affected	v/+	unaffected	NT	unaffected	v/+	-	-	
NGC4	PTPN1 1	c.794G>A; p.(Arg265Gln)	affected	v/+ (de novo)	unaffected	+/+	unaffected	+/+	-	-	
NGC12	SMO	c.1264+3G>C	affected	v/+	affected	v/+	unaffected	NT	Maternal grandmother	affected	v/+
NGC14	BEST1	c.1144G>A; p.(Glu382Lys)	affected	v/+	unaffected	+/+	unaffected	v/+	Paternal grandmother	affected	NT
NGC18	TP63	c.416C>T; p.(Ala139Val)	affected	v/+	affected	v/+	unaffected	+/+	-	-	-
	PKD2	c.1094+1G>A	affected	v/+	affected	v/+	unaffected	+/+	-	-	-
Monoallelic families											
NGC16	SMOC 1	c.832C>T; p.(Arg278Cys)	affected	v/+	unaffected	v/+	unaffected	+/+	-	-	-

NT, not tested; v, variant; +, wild-type allele