Table S1. Summary of heterozygous non-synonymous variants.

Gene	Location	Disease association	cDNA change	RefSeq ID	Amino acid change	Predicted function		
						Polyphen-2	SIFT	PROVEAN
Non-synonymous variants that are present in groups 1+2 and absent from group 3 and control data								
HDAC4	2q37.3	Brachydactyly-mental retardation syndrome[a]	c.610G>A	NM_006037	p.G204R	Benign	Tolerated	Neutral
AGBL2	11p11.2		c.2135A>C	NM_02478	p.D712A	Damaging	Tolerated	Deleterious
SLC15A3	11q12.2		c.269G>T	NM_016582	p.G90V	Benign	Damaging	Deleterious
MRGPRF	11q13.1		c.509T>C	NM_001098515	p.L170P	Damaging	Damaging	Deleterious
CCND1	11q13	Colorectal cancer (susceptibility)[b]	c.826_828del	NM_053056	p.E276del[c]	N.A.	N.A.	Neutral
		von Hippel-Lindau disease (modifier)[b]						
CTTN	11q13		c.1030T>C	NM_001184740	p.R344W	Damaging	Damaging	Deleterious
TIAM1	21q22.1		c.3059C>A	NM_003253	p.S1020Y	Benign	Damaging	Deleterious
CLIC6	21q22.12		c.776_805del	NM_053277	p.V259_S268del[d]	N.A.	N.A.	Neutral
TBX1	22q11.21	DiGeorge syndrome	c.1253delA	NM_080647	p.Y418fsX459	N.A.	N.A.	N.A.
		Velocardiofacial syndrome						
Non-synonymous variants that are present in group 1 and absent from groups 2+3 and control data								
EP400	12q24.33		c.2494C>G	NM_015409	p.R832G	Damaging	Damaging	Deleterious
CEP76	18p11.21		c.1327G>T	NM_024899	p.V443F	Benign	Tolerated	Neutral

N.A.: not applicable.

[a] Caused by heterozygous loss-of-function mutations of *HDAC4*.

[b] Constituted by overexpression of *CCND4*.

[c] This deletion shortens the glutamic acid stretch from nine to eight.

[d] In-frame deletion of 10 amino acids (VEAGVPAGDS).