Supplementary Table S2. Candidate variants at the candidate loci. Novel/rare exonic (except synonymous) and splicing variants were selected, and those with alternative depth <60% of the total depth or not validated by inspection of sequence reads on BamView were filtered out. The strongest candidates for causative mutations are highlighted in pink.

A. In patient 402 in family 1.

chr_name	chr_start	chr_end	ref_base	alt_base	hom_het	snp_quality	tot_depth	alt_depth	region	gene	change
Chromosome 4 rs4689888 (4569460 bp) - rs17779675 (6449514 bp)											
chr04	5755565	5755565	G	A	hom	196	31	31	exonic	EVC	nonsynonymous
Chromosome 18 rs2339102 (23797853 bp) - rs1609839 (24027419 bp)											
chr18	23873494	23873494	C	T	hom	194	85	84	exonic;splicing	TAF4B	stopgain

Chromosome 7 rs4236541 (98830439 bp) - rs28365067 (99272310 bp)
No candidate variants
Chromosome 5 rs186893 (8473747 bp) - rs17225452 (8873310 bp)
No variants because there are no genes in the region.

B. In patient 402 in family 2.

chr_name	chr_start	chr_end	ref_base	alt_base	hom_het	snp_quality	tot_depth	alt_depth	region	gene	change
Chromosome 17 pter to rs3744405 (7193255 bp)											
17	2276785	2276785	G	A	hom	105	80	80	exonic	SGSM2	nonsynonymous
17	4647504	4647507	AAAC	-	hom	7893	112	101	exonic	ZMYND15	frameshift del
17	4863328	4863328	G	A	hom	195	58	58	exonic	SPAG7	nonsynonymous

Chromosome 14 rs8010057 (23506110 bp) - rs2332572 (26341293 bp)
No candidate variants
Chromosome 7 rs4621714 (153356432 bp) - rs10229774 (153652188 bp)
No candidate variants

PloyPhen score : Ranges from 0 to 1, and 0 is benign and 1 is damaging.
MutPred: Score is the probability that the mutation is deleterious.
SIFT score ranges from 0 to 1. Amino acid substitution is predicted damaging if the score is ≤ 0.05 and tolerated if the score is ≥ 0.05.
Mutation Taster: The probability value refers to the prediction, i.e. a value close to 1 indicates a high 'security' of the prediction.

dbSNP132	freq	Sanger Seq	Phenotype	PolyPhen	MutPred	SIFT Score	Mutation Taster
rs141859946	0.001	not done	EVC	BENIGN, 0.123	BENIGN, p= 0.309	TOLERATED, 0.71	BENIGN, p=0.99
(novel)	.	validated	-				

dbSNP132	freq	Sanger Seq	Phenotype	PolyPhen	MutPred	SIFT Score	Mutation Taster
rs140107244	0.00	not done	-	BENIGN, 0.121	BENIGN, p = 0.352	TOLERATED, 0.59	DISEASE CAUSING, p=0.99
(novel)	-	validated	-				
rs201256045	NA	validated	-	BENIGN, 0.013	BENIGN, p = 0.252	DAMAGING, 0.01	DISEASE CAUSING, p=0.99