Supp. Table S1. Rare, heterozygous, nonsynonymous sequence variants present in all four affected individuals who underwent exome sequencing

Chr	Nucleotide (hg19)	Gene Symbol and Accession Number	Variant[a]	Amino Acid change	Gene Name
2	179440229	TTN (NM_001267550.1)	c.70630A>G	p.I23544V	Titin
2	216279562	FN1 (NM_212482.1)	c.1939C>T	p.P647S	Fibronectin 1
7	27140773	HOXA2 (NM_006735.3)	c.703C>T[b]	p.Q235*	Homeobox A2
11	108060036	NPAT (NM_002519.2)	c.353C>T	p.A118V	Nuclear protein, ataxia-telangiectasia locus
11	118958999	HMBS (NM__000190.3)	c.68T>C	p.V23A	Hydroxymethylbilane synthase
17	79914802	NOTUM (NM_178493.5)	c.844A>G	p.T282A	Notum pectinacetylesterase homolog
22	24580811	SUSD2 (NM_019601.3)	c.693_698delTTTCAC	p.F232_T233del	Sushi domain containing 2

[a]Nucleotide numbering reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence

[b]The HOXA2 nonsense variant has been submitted to the LOVD HOX2 locus specific database at http://www.lovd.nl/HOXA2