Table S1 Variant prioritization and filtering strategy used to analyze exome sequencing data

Total called variants	41,308
Rare variants (< 1%)[a]	1,918
Read number (> 20)	1,434
Excluding intergenic and intronic variants	918
Excluding synonymous variants	671
Homozygous variants	84
Shared by all three families[b]	1

[a]Variant frequency equal to or less than 1% in population databases (gnomAD, 1000 Genomes Project, ExAC, and our in-house data of 377 exomes).
[b]Known variant, in concordance with the observed phenotype.

Table S2 Regions of homozygosity by descent in probands of all three families, homozygous block (colored) around the *ISCA1* variant locus (green), identified by exome sequencing data from family 1 and family 2 (reported earlier) and family 3 (present family) demonstrates that the *ISCA1* mutation (green) is due to a founder effect

Shukla et al[4]								Present study			
Family 1				Family 2				Family 3			
Chr	Genomic position	Variant	Zygosity	Chr	Genomic position	Variant	Zygosity	Chr	Genomic position	Variant	Zygosity
chr9	82267732	G	Hom								
chr9	82286286	G	Hom								
chr9	82320887	T	Hom	chr9	82320887	T					
								chr9	82323665	T	Hom
chr9	84205860	A	Hom	chr9	84205860	A	Hom	chr9	84205860	A	Hom
				chr9	84214974	G	Hom	chr9	84214974	G	Hom
chr9	84267147	C	Hom	chr9	84267147	C	Hom	chr9	84267147	C	Hom
				chr9	84268878	G	Hom	chr9	84268878	G	Hom
chr9	85613354	G	Hom	chr9	85613354	G	Hom	chr9	85613354	G	Hom
chr9	85677562	A	Hom	chr9	85677562	A	Hom	chr9	85677562	A	Hom
chr9	85964563	T	Hom	chr9	85964563	T	Hom	chr9	85964563	T	Hom
chr9	85987880	–	Hom	chr9	85987880	–	Hom	chr9	85987880	–	Hom
chr9	86153159	G	Hom	chr9	86153159	G	Hom	chr9	86153159	G	Hom
chr9	86238086	C	Hom	chr9	86238086	C	Hom	chr9	86238086	C	Hom
chr9	86241489	G	Hom	chr9	86241489	G	Hom	chr9	86241489	G	Hom
chr9	86278817	A	Hom	chr9	86278817	A	Hom	chr9	86278817	A	Hom
chr9	86284178	A	Hom	chr9	86284178	A	Hom	chr9	86284178	A	Hom
chr9	86468715	T	Hom	chr9	86468715	T	Hom	chr9	86468715	T	Hom
chr9	86504005	T	Hom	chr9	86504005	T	Hom	chr9	86504005	T	Hom
chr9	86592026	G	Hom	chr9	86592026	G	Hom	chr9	86592026	G	Hom
chr9	87285915	T	Hom	chr9	87285915	T	Hom	chr9	87285915	T	Hom
chr9	88694179	T	Hom	chr9	88694179	T	Hom	chr9	88694179	T	Hom
chr9	**88881089**	**T**	**Hom**	**chr9**	**88881089**	**T**	**Hom**	**chr9**	**88881089**	**T**	**Hom**
chr9	88924057	C	Hom	chr9	88924057	C	Hom	chr9	88924057	C	Hom
chr9	88925774	G	Hom	chr9	88925774	G	Hom	chr9	90062823	G	Hom
chr9	90258248	T	Hom								

Table S2 (*Continued*)

Shukla et al[4]								Present study			
Family 1				Family 2				Family 3			
Chr	Genomic position	Variant	Zygosity	Chr	Genomic position	Variant	Zygosity	Chr	Genomic position	Variant	Zygosity
chr9	90258413	C	Hom								
chr9	90258413	–	Hom								
chr9	90258415	C	Hom								
				chr9	94518328	T	Hom				
chr9	95219597	A	Hom	chr9	95219597	A	Hom				
chr9	95738781	A	Hom	chr9	95766471	T	Hom				
chr9	95780465	G	Hom	chr9	95780465	G	Hom				
				chr9	95838070	C	Hom				
				chr9	95840077	G	Hom				
				chr9	95840256	G	Hom				

Abbreviation: Chr, chromosome.