S1 Table. Filtering process for gene variants identified through whole exome sequencing

	MS-II-1	AL-IV-3	BG-II-1
Total number of homozygous coding variants or within 20 bp of intron-exon boundaries with MAF <0.5% in Exac, 1000 genomes and eps6500	44 variants (see Table S4 for details)	26 variants (see Table S2 for details)	23 variants (see Table S3 for details)
Of which stop, frameshift or consensus splice variants	AGAP3: rs766984834, c.94_95insGGGG:p.C32fs MNS1: rs185005213, MNS1:NM_018365:c.C724T:p.R242X MST1L: rs200532237, NM_001271733:c.811dupG:p.A271fs SLAIN1: rs201380414, NM_001242868:c.219_220insGG:p.A73fs	MNS1: rs185005213, MNS1:NM_018365:p.R242X	MNS1: rs185005213, MNS1:NM_018365:p.R242X MYO3A:NM_017433:p.T26fs VEZF1:NM_007146:p.Q349QQQ UBXN11: NM_001077262.1:p.GPGPSPCPG506G
Of which (associated with left-right body axis establishment in the literature) segregated with the phenotype in the family	MNS1: rs185005213, MNS1:NM_018365:c.C724T:p.R242X	MNS1: rs185005213, MNS1:NM_018365:p.R242X	MNS1: rs185005213, MNS1:NM_018365:p.R242X

MAF: Minor allele frequency