Supplementary Table S1

BICD2 variant	Classification (ACMG criteria)	Prediction programs	conservation	Frequency in GnomAD	Reported in ClinVar database (number of entries)	Reported in the literature
p1 c.320C>T, p.(Ser107Leu) rs398123028	Pathogenic (PM1, PM2_sup, PP1, PS3, PS4_mod)	Mostly predicted as pathogenic: MutationTaster: disease causing PROVEAN: damaging SIFT: damaging Revel: 0.36 (uncertain)	Strong	-	ClinVar: pathogenic (8)	Bansagi et al., 2017 Unger et al., 2016 Rossor et al., 2015 Neveling et al., 2013 Peeters et al., 2013 Oates et al., 2013
p2 c.1195C>T, p.(Arg399Cys) rs200542458	Uncertain significance (BP4_sup, PM2_sup)	Conflicting predictions: MutationTaster: disease causing PROVEAN: neutral SIFT: tolerated Revel: 0.21	Moderate	0.0000159	ClinVar: uncertain significance (1), likely benign (1)	Not reported in literature
p3 and p4 c.2189G>A, p.(Arg730His) rs943974428	Uncertain significance (PM2_sup, PM1)	Mostly predicted as pathogenic: MutationTaster: disease causing PROVEAN: damaging SIFT: damaging Revel: 0.631	Strong	-	Not reported in ClinVar	Not reported in literature
p5 and p6 c.1904G>T, p.(Arg635Leu)	Uncertain significance (PM2_sup)	Mostly predicted as pathogenic: MutationTaster: disease causing PROVEAN: damaging SIFT: damaging Revel: 0.525	Strong	-	Not reported in ClinVar	Not reported in literature
p7 c.2452A>G, p.(Lys818Glu) rs200341779	Uncertain significance (BP4_mod)	Conflicting predictions: MutationTaster: disease causing PROVEAN: neutral SIFT: damaging Revel: 0.112	Moderate	0.0000322	ClinVar: likely benign (2)	Not reported in literature

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Variants in other genes	**Classification**	**Prediction programs**	**conservation**	**Frequency in GnomAD**	**Reported in ClinVar database**	**Reported in the literature**
p5 and p6 *FLNC* c.2272G>A, p.(Val758Met) rs371418145	Uncertain significance	Mostly predicted as pathogenic: Mutationtaster: disease causing PROVEAN: damaging SIFT: damaging Revel: 0.506	moderate (conservation score low)	0.0000725	ClinVar: uncertain significance (5)	Janin et al., 2017 (supplement)
p7 *COL6A1* c.1694G>A, p.(Arg565Gln) rs886057155	Uncertain significance (BP4_mod)	Mostly predicted as benign: Mutationtaster: polymorphism PROVEAN: neutral SIFT: tolerated Revel: 0.175	weak	0.000026	ClinVar: uncertain significance (4), benign (1)	Not reported in literature