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Supplemental Data

Mutations in *BICD2*, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy

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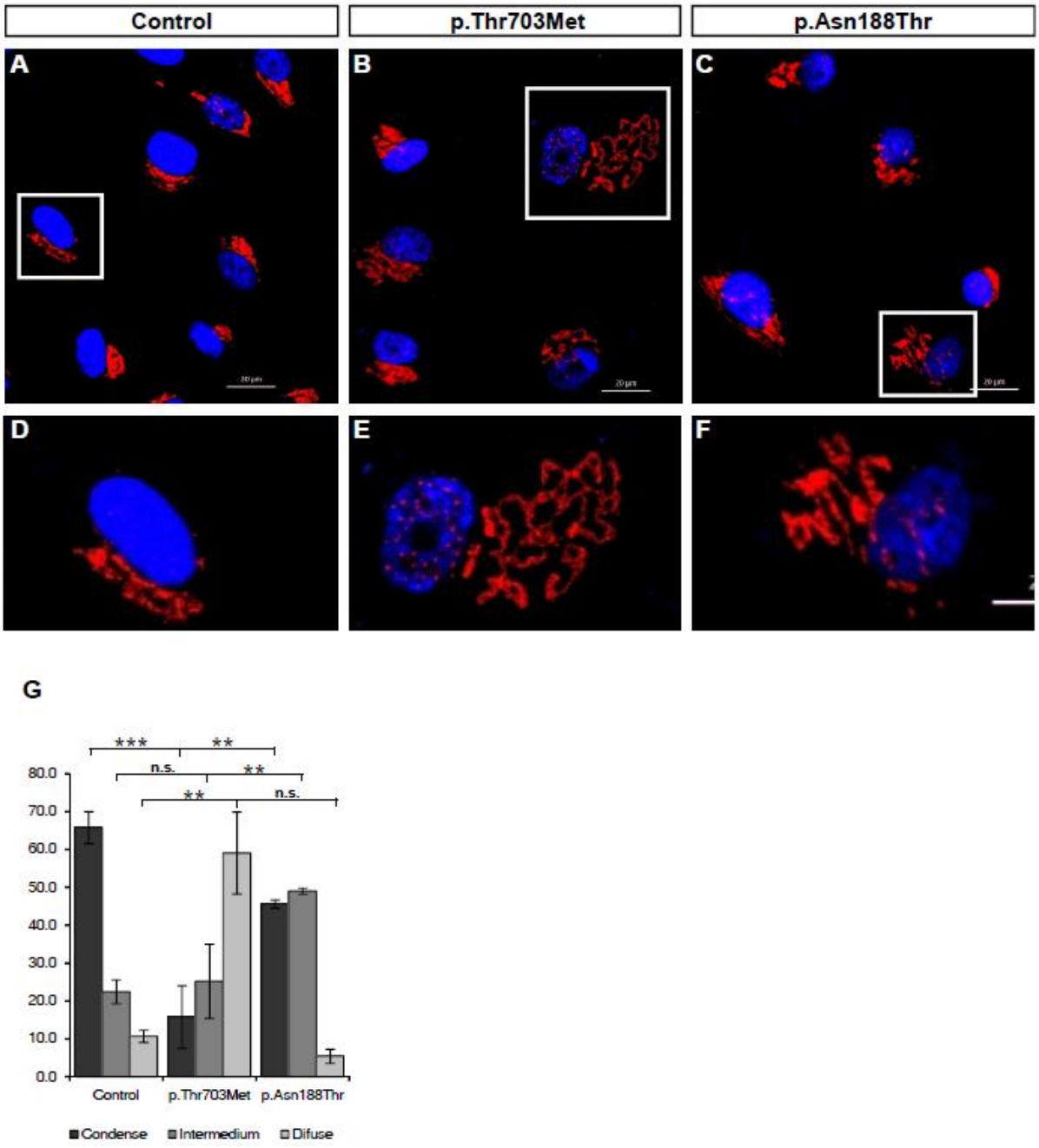

Figure S1. Mutations in *BICD2* Cause Severe Golgi Fragmentation in Primary Fibroblast Cells

(A–C) Golgi apparatus of control fibroblasts and patients fibroblasts immunostained with antibody against the trans-Golgi marker MG160 (0.0025 mg/ml, Abcam); DNA (blue) is stained with DAPI.

(D–F) Magnified insets of (A–C).

(G) Quantification of Golgi structure defined as condense, intermedium and diffuse in 90 cells of each cell line were counted in three independent experiments and given as mean ± SD. Each quantification was done blindly by two independent persons. Statistical significance is given between controls and each of the two cell lines as *** $p<0.001$; ** $p<0.01$, *$p< 0.05$, n. s. = not significant.

Table S1. Filtering of Annotated Variants

Total variants	27,190
variants on chromosome 9	1046
no known SNPs	118
located in exon or splice site	29
non-synonymous	23
not seen before in other projects	5

Table S2. Detailed Information of Remaining Five Candidate Variants

Chr	Position	Ref	Mut	Reads	Var Reads	% Var Reads	Gene	Ref AA	Mut AA	PhyloP
chr9	21007123	T	G	60	39	65	*PTPLAD2*	-[1]	-[1]	-3.18
chr9	34514466	G	A	146	62	43	*DNAI1*	W	*	5.39
chr9	37707448	A	T	75	29	39	*FRMPD1*	N	I	-0.11
chr9	95491439	G	A	56	25	45	*BICD2*	S	L	3.65
chr9	125621277	T	C	100	56	56	*RC3H2*	M	V	2.39

Chr: chromosome on which the nucleotide change is detected, Position: genomic position at which the nucleotide change is detected, Ref: reference nucleotide at the respective position, Mut: substitution of the nucleotide at the respective position, Read: total number of reads at the respective position, Var reads: number of reads showing the nucleotide substitution, % Var reads: Percentage of reads showing the nucleotide substitution, Gene: Gene in which the nucleotide change is detected, Ref AA: reference amino acid at the respective position, Mut AA: substitution of amino acid resulting from detected nucleotide change, PhyloP: phyloP conservation score for evolutionary conservation. [1]:This variant is located within a splice site, therefore no amino acid is given here.

Table S3. Identified *BICD2* Mutations

Family	Origin	Number of Patients/ Family	gDNA	cDNA	Protein	rs Number	PhyloP Score	SIFT	PolyPhen Score
1	Dutch	7	g.95491439C>T	c.320 C>T	p.Ser107Leu	-	3.65	deleterious	probably damaging 0,99
2	Canadian	2	g.95480229C>T	c.2108C>T	p.Thr703Met	-	5.49	deleterious	probably damaging 1.00
3	Dutch	5	g.95484981A>C	c.563A>C	p.Asn188Thr	-	4.25	deleterious	probably damaging 0.99
4	German	2	g.95491490A>G	c.269A>G	p.Lys90Arg	rs61754130	1.42	tolerated	benign 0.002
5	German	5	g.95491490A>G	c.269A>G	p.Lys90Arg	rs61754130	1.42	tolerated	benign 0.002

Family: numbering of the investigated families, Origin: nationalities of the respective families, gDNA: genomic position of detected variant in BICD2, cDNA: cDNA position of detected variant in BICD2, Protein: detected mutation on protein level, rs number: SNP number as given by dbSNP database, SIFT/PolyPhen: Prediction programs used for determination of pathogenicity, PhyloP: PhyloP conservation score for evolutionary conservation.