Table S4. Pathogenic variants in *NDP* gene

Nucleotide change	Amino acid change	N. of families identified in this study	Minor allele frequency			*In-silico* prediction						Conserved domain	Associated phenotype	Known variant [reported in our earlier study]	Pathogenicity (evidenced criteria)*
			gnomAD [27]	HGVD [28]	TOMMO 3.5kJPN v2 [29]	GERP++ [33]	SIFT [34]	M-CAP [35]	REVEL [36]	Polyphen2 HumDIV [37]	CADD [38]				
c.11_12del	p.H4Rfs*21	1	NL	NL	NL	NL	NA	NA	NA	NA	NA	NA	ND	Yes	Pathogenic (PVS1+PS1+PM2)
c.53T>A	p.I18K	1	NL	NL	NL	5.810	0.030	0.714	0.670	0.008	23.4	signal sequence	FEVR(DI)	[23]	Likely pathogenic (PM1+PM2+PP2+PP3)
c.88_104del	p.F30Pfs*21	1	NL	NL	NL	NL	NA	NA	NA	NA	NA	NA	ND	No	Likely pathogenic (PVS1+PM2)
c.112C>T	p.R38C	1	NL	NL	NL	5.810	0.000	0.968	0.730	0.999	28.3	NA	ND	Yes	Likely pathogenic (PS1+PM2+PP2+PP3)
c.162G>C	p.K54N	2	NL	NL	NL	2.820	0.220	0.122	0.520	0.999	23.1	(CDD: 153492)	FEVR(XL)	[23]	Pathogenic (PS1+PS3+PM1+PM2+PP2+PP3)
c.175-1G>A	-	1	NL	NL	NL	5.960	NA	NA	NA	NA	35.0	NA	ND	[23]	Pathogenic (PVS1+PM2+PP3)
c.194G>A	p.C65Y	1	NL	NL	NL	5.960	0.000	0.862	0.916	0.997	29.3	(CDD: 153492)	ND	[23]	Pathogenic (PS1+PM1+PM2+PM5+PP2+PP3)
c.290G>C	p.R97P	1	NL	NL	NL	5.960	0.000	0.893	0.841	0.999	32.0	(CDD: 153492)	ND	[23]	Pathogenic (PS1+PM1+PM2+PM5+PP2+PP3)
c.295_300del	p.Q99_T100del	1	NL	NL	NL	NL	NA	NA	NA	NA	NA	(CDD: 153492)	ND	No	Likely pathogenic (PM1+PM2+PM4)
c.334_340del	p.G112Cfs*148	1	NL	NL	NL	NL	NA	NA	NA	NA	NA	NA	ND	No	Likely pathogenic

															(PVS1+PM2)
c.344G>T	p.R115L	2	NL	NL	NL	5.960	0.050	0.882	0.784	0.997	26.7	(CDD: 153492)	FEVR(XL/SP)	[23]	Pathogenic (PS3+PM1+PM2+PM5+PP2+PP3)
c.376T>G	p.C126G	1	NL	NL	NL	5.960	0.000	0.847	0.886	0.997	26.8	(CDD: 153492)	ND	No	Likely pathogenic (PM1+PM2+PM5+PP2+PP3)

Underlined values are classified as "not deleterious" according to the cutoff values ([33]-[37]); CDD, Conserved Domain Database (http://www.ncbi.nlm.nih.gov/RefSeq/, [31]); DI, digenic inheritance; FEVR, familial exudative vitreoretinopathy; NA, not applicable; ND, Norrie disease; NL, not listed; SP, sporadic; XL, X-llinked.

* Based on the recommendation of the American College of Medical Genetics and Genomics (ACMG) standard and guidelines [32].