Table 1. List of *CHRDL1* variants identified in ten families with X-linked megalocornea

Family	Origin	DNA level	Predicted at protein level	ACMG criteria	ACMG classification	Reference
1	Vietnamese	c.301+2T>G	p.?	PVS1, PS1, PM2, PP4	Pathogenic	(Webb et al. 2012)
2	White Czech	c.1123C>T	p.(Gln375*)	PVS1, PM2, PP4	Pathogenic	Novel
3	White Czech	c.976A>T	p.(Lys326*)	PVS1, PM2, PP4	Pathogenic	Novel
4	White Czech	c.94+1G>A	p.?	PVS1, PP1, PM2, PP4	Pathogenic	Novel
5	White Czech	c.207G>C	p.(Glu69Asp)	PM2, PP3, PP4	Variant of uncertain significance	Novel
6	White New Zealander	c.483dup	p.(Lys162Glnfs*30)	PVS1, PP1, PM2, PP4	Pathogenic	Novel
7	White British	c.436T>G	p.(Cys146Gly)	PM1, PM2, PP3, PP4	Likely pathogenic	Novel
8	North African	c.1156+1G>T	p.?	PVS1, PM2, PP4	Pathogenic	Novel
9	White Australian	c.229C>T	p.(Arg77*)	PVS1, PS1, PM2, PP4	Pathogenic	(Davidson et al. 2014)
10	White British	c.968G>T	p.(Cys323Phe)	PM1, PM2, PP3, PP4	Likely pathogenic	Novel

NM_001143981.2 was taken as the reference sequence.

All variants are absent from gnomAD v.2.1.1 providing allele frequencies of 125,748 exomes and 15,708 genomes from unrelated individuals.