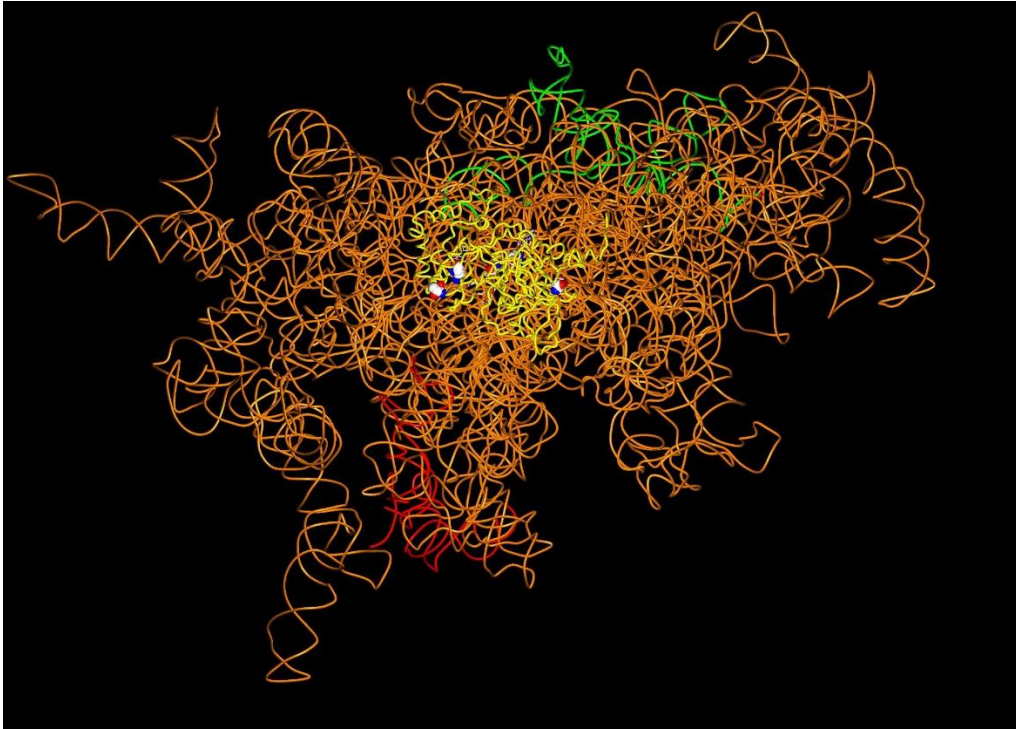


Supplementary Material

Fig. S1. Homology model of RPL3L binding to the ribosome.



RPL3L and ribosomal RNA are shown in worm representation with RPL3L in yellow, the 28S RNA in orange, the 5S RNA in red, and the 5.8S RNA in green. Residues in RPL3L with significant variants are shown in sphere representation and colored by atom type.

Table S1. M-CAP scores and *in silico* prediction of the *RPL3L* variants identified in this study.

Family	Genomic location (hg19)	HGVS cDNA	HGVS protein	M-CAP scores	M-CAP prediction
1	Chr16:1996654	c.923A>T	p.(Asp308Val)	0.061	Possibly Pathogenic
	Chr16:1995856	c.1027C>T	p.(Arg343Trp)	0.054	Possibly Pathogenic
2	Chr16:1997317	c.566C>T	p.(Thr189Met)	0.03	Possibly Pathogenic
	Chr16:1996655	c.922G>A	p.(Asp308Asn)	0.044	Possibly Pathogenic
3	Chr16:2000865	c.481C>T	p.(Arg161Trp)	0.064	Possibly Pathogenic
	Chr16:2004073	c.80G>A	p.(Gly27Asp)	0.09	Possibly Pathogenic

Table S2. MPC, MVP2 and PrimateAI prediction scores for the identified *RPL3L* missense variants.

Family	Genomic location (hg19)	HGVS cDNA	HGVS protein	MPC	MVP2	PrimateAI
1	Chr16:1996654	c.923A>T	p.(Asp308Val)	0.63	0.78	0.62
	Chr16:1995856	c.1027C>T	p.(Arg343Trp)	0.54	0.81	0.79
2	Chr16:1997317	c.566C>T	p.(Thr189Met)	0.52	0.25	0.57
	Chr16:1996655	c.922G>A	p.(Asp308Asn)	0.43	0.65	0.67
3	Chr16:2000865	c.481C>T	p.(Arg161Trp)	0.45	0.84	0.79
	Chr16:2004073	c.80G>A	p.(Gly27Asp)	0.55	0.79	0.81