

Supplemental Table 1 – Predicted pathogenicity of identified *CEP120* variants

	SIFT	Polyphen-2	Provean	MutationAssessor	dbSNP	EVS	ExAC
p.Val194Ala	0	0,999	Deleterious (-2.98)	M (2.32)	no	no	no
p.Ala199Pro	0	1	Deleterious (-3.40)	M (2.36)	rs367600930	0.024/0.0/0.0162 (no hom)	0.0041% (no hom)
p.Leu712Phe	0,1*	0,997	Deleterious (-3.37)	M (2.48)	rs114280473 (MAF 0.0018)	0.4302/0.1362/0.3306 (no hom)	0.003547 (no hom)
p.Leu726Pro	0	0,999	Deleterious (-5.11)	M (2.46)	no	No	no
p.Ile975Ser	0	0,998	Deleterious (-4.28)	L (1.89)	no	No	no

Legend: MAF: minor allele frequency; hom: homozygotes. Range and pathogenicity scores for each software are as follows: SIFT: range 0-1 (deleterious ≤ 0.05 ; tolerated > 0.05); PolyPhen-2: range 0-1 (probably damaging ≥ 0.957 ; possibly damaging $0.453 \geq \text{score} \leq 0.957$; benign ≤ 0.453); Provean: range -13 - +4 (deleterious ≤ -2.5 ; neutral > -2.5); Mutation Assessor: range -5.76 - +5.37 (neutral < 0.6 ; low impact $0.6 \geq \text{score} < 2.0$, medium impact $2.0 \geq \text{score} < 3.7$, high impact ≥ 3.7).