Supplementary Table SIII **Exonic and splice site variants identified by whole-exome sequencing within the premature ovarian failure (POF) associated deletion regions.**

Case ID	Gene	RefSeq	Chr	Position (hg19)	rsID	Zygosity	cDNA	Protein	Allele frequency in ExAC (%)[a]	Effect of mutation	Exclusion criteria
Case1	FMN2	NM_020066.4	1	240370629	rs10926166	hom_alt	c.2517C>G	p.(−)	0.3923	Synonymous	NA
Case1	FMN2	NM_020066.4	1	240371106	rs11586155	het	c.2994C>T	p.(−)	0.3577	Synonymous	Likely a sequencing error
Case1	FMN2	NM_020066.4	1	240371211	rs201587786	hom_alt	c.3099G>T	p.(−)	NA	Synonymous	NA
Case1	FMN2	NM_020066.4	1	240371554	rs12732924	hom_alt	c.3442A>G	p.(−)	0.9111	Missense	NA
Case7	CYP2E1	NM_000773.3	10	135347397	rs915909	hom_alt	c.963T>C	p.(−)	0.9999	Synonymous	NA
Case8	CYP2E1	NM_000773.3	10	135347397	rs915909	hom_alt	c.963T>C	p.(−)	0.9999	Synonymous	NA
Case8	CYP2E1	NM_000773.3	10	135351362	rs2515641	hom_alt	c.1263T>C	p.(−)	0.8869	Synonymous	NA
Case10	CPEB1	NM_001079533.1	15	83224696	rs1145171	hom_alt	c.331G>A	p.Val111Ile	1	Missense	NA
Case10	AP3B2	NM_004644.4	15	83330605	rs376398184	hom_alt	c.2931G>A	p.Met977Ile	0.0001117	Missense	NA
Case10	AP3B2	NM_004644.4	15	83346018	rs3217364	het	c.1488+7delG	p.(−)	0.007189	Splice site	Homozygous genotype validated by Sanger sequencing
Case10	FSD2	NM_001007122.3	15	83428193	rs1108135	hom_alt	c.2157C>T	p.(−)	0.1091	Synonymous	NA
Case10	WHAMM	NM_001080435.2	15	83502065	rs11259953	hom_alt	c.2207A>C	p.His736Pro	0.102	Missense	NA
Case10	WHAMM	NM_001080435.2	15	83502066	rs11259954	hom_alt	c.2208C>G	p.His736Gln	0.102	Missense	NA
Case10	C15orf40	NM_001160116.1	15	83680287	rs4842860	hom_alt	c.73T>C	p.Cys25Arg	0.6396	Missense	NA
Case10	ADAMTSL3	NM_207517.2	15	84488636	rs4483821	hom_alt	c.437A>G	p.His146Arg	0.4528	Missense	NA
Case10	ADAMTSL3	NM_207517.2	15	84539619	rs4144691	hom_alt	c.868C>G	p.Leu290Val	0.8036	Missense	NA
Case10	ADAMTSL3	NM_207517.2	15	84581904	rs4842923	hom_alt	c.1761T>C	p.(−)	0.522	Synonymous	NA
Case10	ADAMTSL3	NM_207517.2	15	84582124	rs4842838	hom_alt	c.1981G>T	p.Val661Leu	0.5218	Missense	NA
Case10	ADAMTSL3	NM_207517.2	15	84651185	rs8031704	hom_alt	c.2805C>A	p.(−)	0.3301	Synonymous	NA
Case10	ADAMTSL3	NM_207517.2	15	84651290	rs7176737	hom_alt	c.2910T>C	p.(−)	0.776	Synonymous	NA
Case10	ADAMTSL3	NM_207517.2	15	84706461	rs950169	hom_alt	c.4979C>T	p.Thr1660Ile	0.2763	Missense	NA
Case13	TTLL1	NM_012263.4	22	43455531	rs1132079	hom_alt	c.750G>A	p.(−)	0.308	Splice site	NA
Case13	TTLL1	NM_012263.4	22	43459846	rs1052160	hom_alt	c.720C>T	p.(−)	0.2977	Synonymous	NA

NA, not available; het, heterozygous; hom_alt, homozygous variant for alternative allele

[a]The allele frequency of variants present in the ExAC dataset are shown in Europeans (Non-Finnish).