

Supplementary

Table S1 Detailed information of the variants detected in the 11 fetuses

ID	CHROM	POS	ID	REF	ALT	Func.refGene	Related_gene	GeneDetail.refGene	ExonicFunc.refGene	AAChange.refGene	ExAC_ALL	gnomAD_AF	SIFT_pred	Polyphen2_HDIV_pred	MetaSVM_pred	Hom/Het
CPAM1	chr8	71299070	.	T	C	exonic	EYA1	.	nonsynonymous SNV	EYA1:NM_172060:exon7:c.A704G;p.Q235R,EYA1:NM_172058:exon8:c.A803G;p.Q268R,EYA1:NM_172059:exon8:c.A788G;p.Q263R,EYA1:NM_000503:exon9:c.A803G;p.Q268R,EYA1:NM_001288574:exon9:c.A785G;p.Q262R,EYA1:NM_001288575:exon9:c.A437G;p.Q146R	.	0	T	D	D	Het
CPAM2	chr1	146074349	.	T	G	exonic	NBPF10	.	nonsynonymous SNV	NBPF10:NM_001039703:exon76:c.A9493C;p.T3165P,NBPF10:NM_001302371:exon80:c.A10000C;p.T3334P	.	0	.	.	.	Hom
CPAM2	chr13	32770441	.	CA	C	exonic	PDS5B	.	frameshift deletion	PDS5B:NM_015032:exon32:c.3946delA;p.K1318Nfs*76	0.0007	7.168e-05	.	.	.	Het
CPAM3	chr7	100955628	.	T	TG	exonic	MUC3A	.	frameshift insertion	MUC3A:NM_005960:exon2:c.3849_3850insG;p.T1284Dfs*36	.	0	.	.	.	Het
CPAM3	chr22	38843700	.	CG	C	exonic	NPTXR	.	frameshift deletion	NPTXR:NM_014293:exon1:c.158delC;p.P53Rfs*7	.	0	.	.	.	Het
CPAM4	chr1	16589962	.	C	T	exonic	NBPF1	.	unknown	UNKNOWN	5.952e-05	3.696e-05	.	.	.	Het
CPAM4	chr2	241602972	.	G	T	exonic	THAP4	.	nonsynonymous SNV	THAP4:NM_001164356:exon3:c.C272A;p.T91K,THAP4:NM_015963:exon4:c.C1508A;p.T503K	.	0	T	D	D	Het
CPAM4	chr7	2844151	.	AC	A	exonic	GNA12	.	frameshift deletion	GNA12:NM_001293092:exon1:c.10delG;p.V4Wfs*55,GNA12:NM_007353:exon1:c.10delG;p.V4Wfs*55	.	0	.	.	.	Het
CPAM5	chr1	149005125	.	C	T	exonic	PDE4DIP	.	nonsynonymous SNV	PDE4DIP:NM_001350520:exon23:c.C4589T;p.A1530V,PDE4DIP:NM_001198834:exon27:c.C4103T;p.A1368V,PDE4DIP:NM_014644:exon27:c.C4103T;p.A1368V,PDE4DIP:NM_001198832:exon30:c.C3971T;p.A1324V,PDE4DIP:NM_001350521:exon30:c.C4514T;p.A1505V,PDE4DIP:NM_001350522:exon30:c.C4301T;p.A1434V,PDE4DIP:NM_001350523:exon30:c.C3968T;p.A1323V	.	0	.	.	.	Het
CPAM5	chr11	1017046	.	GT	G	exonic	MUC6	.	frameshift deletion	MUC6:NM_005961:exon31:c.5754delA;p.P1919Lfs*66	.	0	.	.	.	Het
CPAM7	chr22	26484008	.	GC	G	exonic	SRRD	.	frameshift deletion	SRRD:NM_001013694:exon1:c.119delC;p.A40Gfs*44	.	0	.	.	.	Het
CPAM7	chrX	141906382	.	TCA	T	exonic	MAGEC1	.	frameshift deletion	MAGEC1:NM_005462:exon4:c.979_980del:p.H327Qfs*4	.	0	.	.	.	Hom
CPAM8	chr2	162225499	.	A	C	exonic	FAP	.	nonsynonymous SNV	FAP:NM_001291807:exon4:c.T269G;p.L90W,FAP:NM_004460:exon4:c.T269G;p.L90W	8.36e-06	8.292e-06	D	D	D	Het
CPAM9	chr2	43694467	.	G	T	exonic	PLEKHH2	.	stopgain	PLEKHH2:NM_172069:exon5:c.G373T;p.E125X	.	0	.	.	.	Het
CPAM9	chr9	38395998	.	C	T	exonic	ALDH1B1	.	nonsynonymous SNV	ALDH1B1:NM_000692:exon2:c.C250T;p.R84W	4.96e-05	4.484e-05	D	D	D	Het
CPAM11	chr6	24579941	.	AC	A	exonic	KIAA0319	.	frameshift deletion	KIAA0319:NM_001168376:exon7:c.1153delG;p.V385Sfs*6,KIAA0319:NM_001350404:exon7:c.1270delG;p.V424Sfs*6,KIAA0319:NM_001350405:exon7:c.1189delG;p.V397Sfs*6,KIAA0319:NM_001350406:exon7:c.1153delG;p.V385Sfs*6,KIAA0319:NM_001168375:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_001168377:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_001350403:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_001350407:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_001350408:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_014809:exon8:c.1288delG;p.V430Sfs*6,KIAA0319:NM_001168374:exon9:c.1261delG;p.V421Sfs*6,KIAA0319:NM_001350409:exon9:c.832delG;p.V278Sfs*6,KIAA0319:NM_001350410:exon9:c.832delG;p.V278Sfs*6	3.601e-05	1.46e-05	.	.	.	Het
CPAM11	chr7	100958084	.	CCACCTCACACAGTACT	C	exonic	MUC3A	.	frameshift deletion	MUC3A:NM_005960:exon2:c.6306_6321del;p.T2103Pfs*57	.	0	.	.	.	Het
CPAM11	chr9	130681622	.	GCCGCCGCCGCCGCCG	G	exonic	PRDM12	.	frameshift deletion	PRDM12:NM_021619:exon5:c.1058_1073del;p.A354Rfs*109	.	0	.	.	.	Het
CPAM11	chr15	23129938	.	G	C	exonic	GOLGA6L1;GOLGA6L22	.	nonsynonymous SNV	GOLGA6L1:NM_001001413:exon8:c.C1515G;p.H505Q,GOLGA6L22:NM_001271664:exon9:c.C1485G;p.H495Q	0	9.595e-05	.	.	.	Het
CPAM11	chr20	62065486	.	A	AG	exonic	TAF4	.	frameshift insertion	TAF4:NM_003185:exon1:c.324dupC;p.S109Lfs*353	.	0	.	.	.	Hom
CPAM12	chr6	32518555	.	C	G	splicing	HLA-DRB5	NM_002125:exon4:c.763+1G>C	.	.	0	0	.	.	.	Het
CPAM13	chr2	88452479	.	CG	C	exonic	FOXI3	.	frameshift deletion	FOXI3:NM_001135649:exon1:c.56delC;p.P19Rfs*122	.	0	.	.	.	Het
CPAM13	chr7	100955615	.	C	CAA	exonic	MUC3A	.	frameshift insertion	MUC3A:NM_005960:exon2:c.3836_3837insAA;p.T1280Rfs*65	.	0	.	.	.	Het
CPAM13	chr19	590300	.	CG	C	exonic	HCN2	.	frameshift deletion	HCN2:NM_001194:exon1:c.356delG;p.K122Rfs*140	.	0	.	.	.	Het