Figure S1. Overview of the study procedures. CNV, copy number variation; HLPA, high-throughput ligation-dependent probe amplification; WGS, whole-genome sequencing.

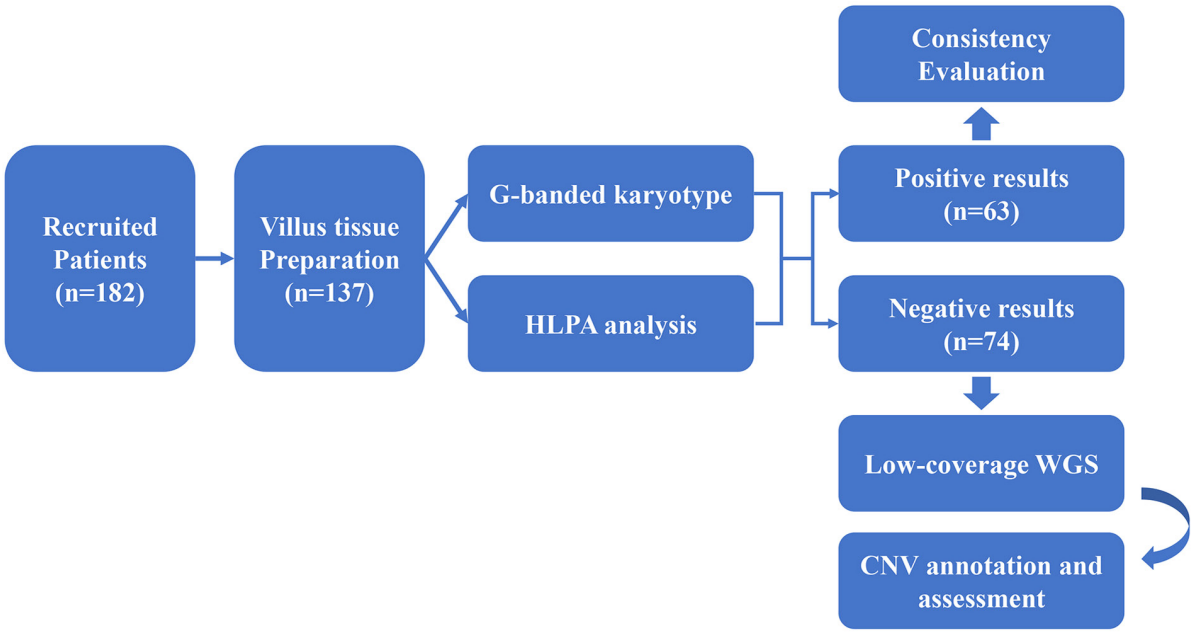

Table SI. Heredity tested by Sanger sequencing of NGS-detected CNVs in abortion tissues.

Case number	CNV location	CNV length (kb)	Duplication/deletion	Genes involved	Heredity of CNVs			Clinical significance
					Paternal	Maternal	*de novo*	
V130	9p24.3	334	Duplication	CBWD1, WASH1, FOXD4, DOCK8, FAM138C, C9orf66, DDX11L5		●		VOUS
V134	3q13.13	504	Duplication	-			●	Suspected benign
	11q13.2	242	Deletion	FAM86C2P		●		VOUS
	15q11.2	203	Deletion	-		●		Suspected benign
V138	2p24.3	811	Duplication	-			●	Suspected benign
V150	4q13.2	105	Deletion	UGT2B28		●		VOUS
	4q31.21	108	Duplication	GYPB, GYPA			●	VOUS
V1001	11p15.1	320	Duplication	TPH1, KCNC1, SERGEF		●		VOUS
	11q13.2	242	Deletion	FAM86C2P			●	VOUS
	12p13.33	376	Duplication	CACNA1C, LRTM2, CACNA2D4, LOC100271702, DCP1B		●		VOUS
V62	10q26.3	128	Deletion	LOC619207, CYP2E1, SYCE1		●		VOUS
	13q22.2	321	Deletion	TBC1D4, COMMD6, UCHL3, LMO7	●			VOUS
V114	4p15.2	191	Duplication	C4orf52		●		VOUS
V133	15q11.2	130	Deletion	PWRN2		●		VOUS
	Xq27.3	202	Deletion	-			●	Suspected benign
V158	7q36.1	308	Duplication	ATP6V0E2-AS1, ZNF862, ATP6V0E2, DPP6			●	VOUS
	7q36.2	183	Duplication	LRP5L, CRYBB2P1, IGLL3P		●		VOUS
	22q11.23q12.1	263	Duplication	-		●		VOUS
V128	5q23.1	519	Duplication	-		●		Suspected benign
	22q11.21	270	Duplication	PRODH, DGCR6, USP18, GGT3P			●	VOUS
	Xp11.23	107	Deletion	ZNF630, SPACA5B, SPACA5, SSX6		●		VOUS
V54	1p11.12	104	Duplication	-	/	/	/	Suspected benign
	16p11.2	236	Duplication	LINC00273	/	/	/	VOUS
	17p13.3	361	Duplication	RAP1GAP2, OR3A1, OR1D2, OR1D5, OR1A1, OR1A2, OR1G1, OR1D4, OR3A2	/	/	/	VOUS
V68	22q11.22	272	Duplication	TOP3B, PPM1F	/	/	/	VOUS
V71	10q23.2	163	Deletion	LOC439994, LOC728190, FAM22A, FAM22D	/	/	/	VOUS
V87	17q21.32	111	Duplication	HOXB-AS3, MIR196A1, MIR10A, HOXB2-9	/	/	/	VOUS
V105	16q24.3	110	Deletion	FANCA	/	/	/	Pathogenic
V113	5p14.3	370	Deletion	INTS4L2, LOC441242, ZNF92	/	/	/	Suspected benign
	7q11.21	542	Deletion	-	/	/	/	VOUS
V77	22q11.23q12.1	263	Deletion	LRP5L, CRYBB2P1, IGLL3P	/	/	/	VOUS
V79	1q31.1	213	Deletion	-	/	/	/	Suspected benign
	11q21q22.1	2.87 Mb	Deletion	JRKL, MAML2, MIR1260B, CCDC82, JRKL-AS1	/	/	/	VOUS
	14q23.2	121	Deletion	SGPP1	/	/	/	VOUS
	15q13.3	519	Duplication	CHRNA7	/	/	/	VOUS

Table SI. Continued.

Case number	CNV location	CNV length (kb)	Duplication/deletion	Genes involved	Heredity of CNVs			Clinical significance
					Paternal	Maternal	*de novo*	
V110	Yp11.32-Yp11.2	7.91 Mb	Deletion	SHOX, PPP2R3B	/	/	/	Pathogenic
V122	4p16.3	3.8 Mb	Deletion	FGFR3, LETM1, WHSC1	/	/	/	Pathogenic
V134	15q25.3	1.3 Mb	Duplication	AGBL1	/	/	/	VOUS

CNV, copy number variation; VOUS, variants of uncertain significance.

Table SII. Detailed information of the cases.

Case number	CNV position	Genes involved	Heredity of CNVs			Clinical significance
			Paternal	Maternal	*de novo*	
V130	9p24.3: 10,001-344,129 (334 kb dup)	CBWD1, WASH1, FOXD4, DOCK8, FAM138C, C9orf66, DDX11L5		●		VOUS
V134	3q13.13: 110,249,785-110,753,998 (504 kb dup)	-			●	Suspected benign
	11q13.2: 67,496,896-67,739,830 (242 kb del)	FAM86C2P		●		VOUS
	15q11.2: 24,583,642-24,786,960 (203 kb del)	-		●		Suspected benign
V138	2p24.3: 3,543,327-14,354,620 (811 kb dup)	-			●	Suspected benign
V150	4q13.2: 70,138,883-70,244,272 (105 kb del)	UGT2B28		●		VOUS
	4q31.21: 144,929,449-145,037,725 (108 kb dup)	GYPB, GYPA			●	VOUS
V1001	11p15.1: 17,760,874-18,081,810 (320 kb dup)	TPH1, KCNC1, SERGEF		●		VOUS
	11q13.2: 67,496,896-67,739,830 (242 kb del)	FAM86C2P			●	VOUS
	12p13.33: 1,937,118-2,313,987 (376 kb dup)	CACNA1C, LRTM2, CACNA2D4, LOC100271702, DCP1B		●		VOUS
V62	10q26.3: 135,247,534-135,376,327 (128 kb del)	LOC619207, CYP2E1, SYCE1		●		VOUS
	13q22.2: 75,964,191-76,285,851 (321 kb del)	TBC1D4, COMMD6, UCHL3, LMO7	●			VOUS
V114	4p15.2: 25,875,906-26,067,557 (191 kb dup)	C4orf52		●		VOUS
V133	15q11.2: 24,359,700-24,489,989 (130 kb del)	PWRN2		●		VOUS
	Xq27.3: 143,134,531-143,337,405 (202 kb del)	-			●	Suspected benign
V158	7q36.1: 149,556,685-149,864,823 (308 kb dup)	ATP6V0E2-AS1, ZNF862, ATP6V0E2, DPP6			●	VOUS
	7q36.2: 153,476,694-153,660,423 (183 kb dup)	LRP5L, CRYBB2P1, IGLL3P		●		VOUS
	22q11.23q12.1: 25,661,151-25,924,924 (263 kb dup)	-		●		VOUS
V128	q23.1 : 120,345,246-120,864,255 (519 kb dup)	-		●		Suspected benign
	22q11.21: 18,644,144-18,914,454 (270 kb dup)	PRODH, DGCR6, USP18, GGT3P			●	VOUS
	Xp11.23: 47,900,955-48,008,640 (107 kb del)	ZNF630, SPACA5B, SPACA5, SSX6		●		VOUS
V54	1p11.12: 48,850,055-48,954,577 (104 kb dup);	-	/	/	/	Suspected benign
	16p11.2: 33,946,471-34,183,059 (236 kb dup);	LINC00273	/	/	/	VOUS
	17p13.3: 2,835,556-3,197,183 (361 kb dup)	RAP1GAP2, OR3A1, OR1D2, OR1D5, OR1A1, OR1A2, OR1G1, OR1D4, OR3A2	/	/	/	VOUS
V68	22q11.22: 22,305,376-22,578,063 (272 kb dup)	TOP3B, PPM1F	/	/	/	VOUS
V71	10q23.2: 88,966,588-89,130,520 (163 kb del)	LOC439994, LOC728190, FAM22A, FAM22D	/	/	/	VOUS
V87	17q21.32: 46,609,588-46,720,700 (111 kb dup)	HOXB-AS3, MIR196A1, MIR10A, HOXB2-9	/	/	/	VOUS
V105	16q24.3 (89820001-89930000) (110 kb del)	FANCA	/	/	/	Pathogenic
V113	5p14.3: 19,083,617-19,454,608 (370 kb del)	INTS4L2, LOC441242, ZNF92	/	/	/	Suspected benign
	7q11.21: 64,581,057-65,123,100 (542 kb del)	-	/	/	/	VOUS
V77	22q11.23q12.1: 25,661,151-25,924,924 (263 kb del)	LRP5L, CRYBB2P1, IGLL3P	/	/	/	VOUS
V79	1q31.1: 189,329,675-189,542,906 (213 kb del)	-	/	/	/	Suspected benign
	11q21q22.1:95,882,557-98,752,576 (2.87 Mb del)	JRKL, MAML2, MIR1260B, CCDC82, JRKL-AS1	/	/	/	VOUS

Table SII. Continued.

Case number	CNV position	Genes involved	Heredity of CNVs			Clinical significance
			Paternal	Maternal	*de novo*	
	14q23.2: 64,177,898-64,299,096 (121 kb del)	SGPP1	/	/	/	VOUS
	15q13.3 :31,994,610-32,514,175 (519 kb dup)	CHRNA7	/	/	/	VOUS
V110	Yp11.32-Yp11.2: 1-7,910,000 (7.91 Mb del)	SHOX, PPP2R3B	/	/	/	Pathogenic
V122	4p16.3: 68,345-3,916,139 (3.8 Mb del)	FGFR3, LETM1, WHSC1	/	/	/	Pathogenic
V134	15q25.3: 86,937,426-88,285,413 (1.3 Mb dup)	AGBL1	/	/	/	VOUS

CNV, copy number variation; VOUS, variants of uncertain significance; del, deletion; dup, duplication.