

Table S1. VCF file comparison of initial analysis and VExP reanalysis.

	Gene	#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	Sample.Id
VCF file from clinical sequencing	<i>RYR1</i>	chr19	38985002	.	G	GCGCTGGGCC	2433.12		*VQSRTTrancheINDEL99.90to100.0(RankSum=0.000;DP=322;ExcessHet=3.9794;FS=29.725;MLEAC=2;MLEAF=0.333;MQ=60.00;MQRankSum=0.000;QD=26.16;ReadPosRankSum=-2.521;SOR=	GT:AD:DP:GQ:PL	0/1:27,9:36:99:1038,0,1260
VExP reanalysis VCF file		19	38985002	.	G	GCGCTGGGCC	682.6	PASS	.270e-01;DP=99;ExcessHet=3.0103;FS=9.764;MLEAC=1;MLEAF=0.500;MQ=60.00;MQRankSum=0.00;QD=14.84;ReadPosRankSum=-4.833e+00;SOR=2.303;	GT:AD:DP:GQ:PL	0/1:27,19:46:99:690,0,1094
VCF file from clinical sequencing	<i>BBS1</i>	chr11	66293652	.	T	G	2038.16		*VQSRTTrancheSNP99.00to99.90 Sum=0.000;DP=278;ExcessHet=3.9794;FS=1.569;MLEAC=2;MLEAF=0.333;MQ=59.58;MQRankSum=-1.991;QD=11.71;ReadPosRankSum=-1.379;SOR=0.574	GT:AD:DP:GQ:PL	0/1:44,45:89:99:1040,0,1682
VExP reanalysis VCF file		11	66293652	.	T	G	2138.49	PASS	kSum=1.97;DP=87;ExcessHet=3.1627;FS=4.430;InbreedingCoeff=-0.0714;MQ=60.00;MQRankSum=0.00;QD=12.43;ReadPosRankSum=0.408;SOR=0.449;VQ:AD:DP:GQ:PGT:PID:PL:99:1/0:66293645 CTCA_C:1098,0,16		

* meas quality metrics failure.