

Sample	Karyotype	GENA TP	IGV TP	GENA FP	Found by GENA	Visible in IGV	Type of error	Comment
Bal1_K	(16)x3, (18q22.1q23)x1,(21q21.3q22.3)x3,(22)x2-3	1,0,1,0	1,1,1,0	chr4 12023045 186077468 1 chr22 1845682 38987003 1	chr16 23006 32047082 3 chr16 52472878 96160593 3 chr21 26422287 45085560 3 chr22 1845682 38987003 1 chr4 12023045 186077468 1	1,1,1,0,1	Methodological and algorithmic errors	Derived from patient with translocation between chromosomes 18 and 21, deletion on 18 chromosome are not found by algorithm but visible in Biop-C data. In initial biopsy little increase of coverage on chromosome 22, we observe deletion - mosaicism?
Mar1_K	arr(3)x3,(8)x3[0,62],[16]x2-3	0,0,1	0,0,1		chr16 23006 32047082 3 chr16 52472878 96160593 3	1,1	Methodological errors	Low quality of input data
Sav4_K	arr(16,22)x3	0,1	0,1	chr16 23006 32047082 3	chr16 23006 32047082 3	1,1	CNVs on the correct chromosome but with different coordinates	
Shen2_e	arr[hg38](11q13.2q25)x1	0	0	chr11 72083007 135123187 3	chr22 10853537 51293848 3 chr11 72083007 135123187 3	1	Strong evidence of mosaicism	
Shen2_K	arr[hg38](11q13.2q25)x1	0	0				Strong evidence of mosaicism	May be an example of reciprocal translocation - initial biopsy with deletion, re-biopsy euploid, whole embryo with duplication
MAD_e3	arr(2)x1	0	0	chr2 4098 28600550 1	chr2 4098 28600550 1	1	CNVs on the correct chromosome but with different coordinates	
Kur3_K	arr(11,12,17,22)x3,(16)x1	0,0,0,0	0,0,0,0				Strong evidence of mosaicism	Derived from patient whose embryos have an increased number of mosaic aneuploidies
Kur3_e	arr(11,12,17,22)x3,(16)x1	0,0,0,0	0,0,0,0				Strong evidence of mosaicism	Derived from patient whose embryos have an increased number of mosaic aneuploidies
6069_plus	seq[GRCh37](1p36.33p13.2)x1,(1p13.2q21.1)x3,(22)x3	0,0,0	0,0,0				Methodological errors	
Pet1_K	arr[hg38](3p21.31q29)x1,(15)x3	0,0	0,0				Methodological errors	
Kov1_e	arr[hg19](1,8)x3,(2p25.1-p21)x3	1,0,0	1,0,0		chr1 259078 247762120 3	1	Methodological errors	
Vla1_K_0705	arr[hg38](6p25.1q16.1)x3,(18q22.2q23)x1	0,0	0,0				Methodological errors	Derived from patient with complex rearrangement between chromosomes 6, 18 and 21
Sach2_K	arr(13,22)x1	0,1	1,1		chr22 1845682 33275708 1	1	Algorithmic errors	
Boc_e4	seq(18)x1	0	0				Strong evidence of mosaicism	Initial and re-biopsies have 18 chromosome aneuploidy, but whole embryo is euploid
Fisher1_K	arr(15)x3	0	0				Methodological errors	
Sav4_e8	arr(16,22)x3	1,0	1,1		chr16 11613466 32047082 3 chr16 52472878 92293666 3	1,1	Algorithmic errors	
6069_1_K	seq[GRCh37] (11p11.12q12.2)x3,(18)x1,(22)x3	0,1,1	0,1,1		chr18 221524 80536469 1 chr22 10853537 51293848 3	1,1	CNVs in pericentromeric regions	
Shur1_K_2205	seq[hg19](10q25.1q26.11)x3	0	0				Methodological errors	