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SHORT COMMUNICATION

The Discordance between G-Banding Karyotyping and Microarray in Structural Abnormality

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SUMMARY

Background: **Cytomolecular genetic laboratory techniques have developed from conventional G-banding karyotyping to whole genome sequencing. Although resolution has greatly increased, various cytogenetic techniques have their advantages and limitations in detecting genomic variations.**
Methods: **We compared the chromosomal abnormalities detected by G-banding karyotyping and SNP-based microarray testing in 62 patients from July 2020 to December 2022. We analyzed their difference according to chromosomal abnormalities, including numerical and structural and others.**
Results: **Of the 62 patients, 28 patients showed chromosomal aberration detected in one or more of the two test methods. Aneuploidy was detected in both methods, while gain and loss less than 3 Mb were only detectable by the microarray. G-banding karyotyping is fundamental to detect structural chromosome rearrangement such as inversions, ring chromosomes, and translocations, but additional breakpoint or unknown origin materials information obtained from microarray. Loss of heterozygosity was only detectable in microarray, and mosaicism had limitations in both G-banding karyotyping and microarray.**
Conclusions: **Various disease cause genomic structural variants, it is very important to detect this. We showed discordance between G-banding karyotyping and SNP based microarray in clinical laboratory. It can be helpful to clinical physicians to decide which diagnostic tool to use.**
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Supplementary Data

Table S1. Summary of 28 discrepancy results between G-banding karyotyping and microarray.

Category	Abnormality type	N	G-banding karyotyping	Microarray	Size
Numerical	Aneuploidy	P1	47, XX, +21	arr (21) x 3	
Structural	Loss	P2	46, XX	arr 15q11.2 (22,770,421 - 23,282,799) x 1	512 kb
		P3	46, XX	arr 7q11.23 (72,718,277 - 74,141,603) x 1	1.4 Mb
		P4	46, XX	arr 17p12 (14,087,933 - 15,484,358) x 1	1.4 Mb
		P5	46, XY	arr 1q21.1q21.2 (146,105,170 - 147,830,830) x 1	1.7 Mb
		P6	46, XX	arr 2p16.3 (50,307,926 - 50,469,800) x 1	161 Kb
		P7	46, XX	arr 15q11.2q13.1 (23,615,768 - 28,828,168) x 1	5.2 Mb
	Gain	P8	46, XY	arr 1p32.3 (54,898,830 - 55,347,872) x 3	449 kb
		P9	46, XX	arr Xq28 (154,110,363 - 154,568,758) x 3	458 kb
		P10	46, XY	arr 8p23.2 (3,686,943 - 5,951,565) x 3	2.3 Mb
		P11	46, XY	arr 16p13.11p12.3 (15,496,101 - 18,242,713) x 3	2.7 Mb
		P12	46, XY	arr 16p11.2 (29,638,640 - 30,177,916) x 3	539 kb
		P13	46, XX	arr 16p11.2 (29,591,078 - 30,177,916) x 3	586 kb
		P14	46, XY	arr 1q21.1q21.2 (145,886,339 - 147,770,397) x 3	1.9 Mb
		P15	46, XY	arr 4p15.2 (25,123,741 - 25,827,716) x 3	704 Kb
	Derivative chromosome	P16	46, XX, der(8)t(8;16)(p23.1;q22)	arr 8p23.3p23.1 (158,048 - 6,999,114) x 1 arr 16q22.3q24.3 (73,752,981 - 90,155,062) x 3	6.8 Mb 16.4 Mb
		P17	46,XY	arr 5p15.2 (11,402,271 - 11,431,656) x 1 arr 15q13.3 (32,003,537 - 32,444,044) x 3	2.9 Kb 440 Kb
	Inversion	P18	46, XX, inv(9)(p12q13)	arr (1 - 22, X) x 2	
		P19	46, XX, inv(9)(p12q13)	arr (1 - 22, X) x 2	
	Additional material unknown	P20	47, XY, +add(9)(q13)	arr 9p24.3p13.1 (203,861 - 38,787,480) x 3 arr 9q32q34.3 (116,890,162 - 141,025,328) x 2 hmz	38.6 Mb 24.1 Mb
	Ring chromosome	P21	46, XX, r(18)	arr 18p11.32 (136,226 - 1,164,159) x 1 arr 18p11.32p11.31 (1,205,753 - 6,112,310) x 3 arr 18q22.2q23 (68,467,799 - 78,014,123) x 1	1.0 Mb 4.9 Kb 9.5 Mb
Other	Loss of Heterozygosity	P20	47, XY, +add(9)(q13)	arr 9q32q34.3 (116,890,162 - 141,025,328) x 2 hmz	24.1 Mb
		P22	46, XX	arr 15q11.2q14 (22,752,398 - 37,494,741) x 2 hmz	14.7 Mb
		P23	46, XY	arr 11p14.3p13 (23,095,870 - 35,511,094) x 2 hmz	12.4 Mb
		P24	46, XY	arr 6p22.2p21.31 (25,410,076 - 36,315,242) x 2 hmz	10.9 Mb
	Mosaicism	P25	47, XXY [13]/46, XY [37]	arr (1 - 22) x 2, (X, Y) x 1	
		P26	47, XX, +psu idic(9)(q12)[3]/ 46, XX [47]	arr (9p) x 2 - 3	
		P27	46, XY, del(16)(q22) [6]/ 46, XY [56]	arr (X, Y) x 1, (1 - 22) x 2	
		P28	45, X [23]/47, XXX [7]	arr (X) x 1 - 2	

Abbreviations: der - derivative chromosome, inv - inversion, add - additional material of unknown origin, r - ring chromosome, psu - pseudo-, idic - isodicentric chromosome, del - deletion, hmz - homozygous.