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The normal karyotype does not exclude genetic problems in fetuses with congenital heart defects

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Introduction: Genetic problems which coexisted with congenital heart defects(CHD) significantly changed the prognosis. Classical karyotype had been performed for years, but it was exchanged by chromosomal microarray (CMA). The aim of this study was to determine the type and frequency of genetic abnormalities in fetuses with diagnosis of CHD using CMA.

Methods: 99 fetuses after detailed fetal echocardiography performed between 2015-2017 in referral fetal cardiology center) were referred to invasive genetic testing. Chromosomal aneuploidies and submicroscopic copy number variations (CNVs) were identified in amniocytes DNA samples using high-resolution CMA.

Results: Out of 99 fetuses with CHD 37(37%) had genetic problems. There were 15 (26%) out of 58 fetuses with an isolated CHD and 22(54%) out of 41 fetuses with CHD and extracardiac malformations. In 16(43%) aneuploidy could be diagnosed by classical karyotype, whereas in 67% only molecular methods gave the final diagnosis.

Category		Number of fetuses	No CNV (%)	Pathogenetic variance		
				Aneuploidy (%)	22q11.2 (%)	Other (%)
Isolated CHD	CTD	21	14	3 (14.3)	3 (14.3)	2 (9.5)
	right heart lesions	5	4 (80)	0	0	1 (20)
	left heart lesions	16	16	0	0	0
	septal defects/AVSD	6	3 (50)	2 (33.3)	0	1 (16.7)
	complex CHD	7	4 (57.1)	0	0	3 (42.9)
	other mild cardiovascular abnormality	3	2 (66.7)	0	1 (33.3)	0
	total	58	43	5 (8.7)	4 (8.6)	6 (10.3)
CHD with extra cardiac defect	CTD	12	6 (50)	2 (16.7)	1 (8.3)	3 (25)
	right heart lesions	2	1 (50)	0	0	1 (50)
	left heart lesions	3	2 (66.7)	0	0	1 (33.3)
	septal defects/AVSD	14	5 (35.7)	6 (42.9)	0	3 (21.4)
	complex CHD	2	0	1 (50)	0	1 (50)
	other mild cardiovascular abnomality	8	5 (62.5)	2 (25)	0	1 (12.5)
	total	41	19 (46.3)	11 (26.8)	1 (2.4)	10 (24.4)
All		99	62 (62.6)	16 (16.1)	5 (5)	16 (16.1)

CTD conotruncal defect; AVSD atrio-ventricular septal defect

Conclusions: Classic karyotype could lead to false diagnosis of isolated CHD. CMA should be considered in all cases with prenatally diagnosed CHD.