GENETIC ANOMALIES ASSOCIATED TO MAJOR CONGENITAL HEART DEFECTS IN A REFERRAL CENTER' SERIES

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OBJECTIVES

To describe the genetic syndromes associated with different types of major CHD studied in BCNatal (Hospital Clínic Barcelona and Hospital Sant Joan de Déu).

METHODS

A retrospective review of the database of BCNatal Fetal Cardiology Unit was conducted. Pregnant women carrying a fetus with major CHD from Jan 2009 to Dec 2021 who agreed to undergo genetic invasive testing, were included. CHD were divided into 5 groups: septal, conotruncal, left heart, right heart and complex defects, according to the anatomical location of the main defect. Regarding genetic testing, conventional karyotype with the addition of FISH22q11 in conotruncal anomalies was performed during the 3 first years of the study. From 2012 onwards, the elective genetic test was array-comparative genomic hybridization (array-CGH). Finally, from 2017, clinical exome sequencing was carried out in CHD associated with extracardiac malformations and normal array-CGH.

RESULTS

	TOTAL	%	Septal	%	Cono truncal	%	Left Heart	%	Right Heart	%	Complex	%
Normal	498	72	186	74	92	69	115	75	53	83	52	62
Aneuploidies	73	11	36	14	8	6	16	10	2	3	11	13
Copy number variations	39	6	8	3	19	14	2	1	3	5	7	8
Monogenic anomalies	17	2	4	2	3	2	2	1	1	2	7	8
Balanced rearrangements	8	1	3	1	1	1	3	2	0	0	1	1
Unbalanced rearrengements	1	0,15	0	0	1	1	0	0	0	0	0	0
Triploidies	2	0,29	0	0	2	2	0	0	0	0	0	0
No result	49	7	16	6	7	5	15	10	5	8	6	7
PATHOLOGICAL	132	19	48	19	33	25	20	13	6	9	25	30
TOTAL	687	100	253	37	133	19	153	22	64	9	84	12

Table 1. Genetic results in every group of CHD.

CONCLUSIONS

In our series of major CHD, association with genetic disorders accounts for 19% of the cases, increasing to 25-30% in conotruncal and complex defects. Genetic testing is crucial to provide accurate counseling to the parents. Array-CGH and clinical exome sequencing provide added value to genetic syndrome diagnosis in CHD.

CHD: Congenital Heart Defect