P-143

Outcome of Congenital Heart Defects Associated with 22q11.2 Deletion

Uzun O. (2), Tucker D. (1), Babaoglu K. (2,3), Massias S. (2), Kumar D. (2), Wilson D.G. (1)
Congenital Anomaly Register and Information Service for Wales (CARIS), Swansea, UK (1); University Hospital of Wales, Cardiff, UK (2); Kocaeli University Medical Faculty, Kocaeli, Turkey (3)

Introduction: 22q11.2 deletion is the most common of the chromosomal microdeletions. This study looks at the association of congenital heart defects with 22q11.2 deletion.
Method: The data used is from the Congenital Anomaly Register and Information Service for Wales (CARIS) from 2001-2010, irrespective of pregnancy outcome.
Results: There were 40 cases of 22q11.2 deletion reported in Wales, giving a gross prevalence of 1.2 per 10,000 total births (1 in 8335 total births). Of these, 30 cases had both 22q11.2 deletion and at least one congenital heart defect (75% of all 22q11.2 deletions reported). In 26 of these 30 cases, pregnancy resulted in a live birth (87%), 3 cases were terminated (10%) and the remaining case was a still birth (3%). Of those with no heart anomaly, 8 were live births (80%), and the remaining 2 cases were terminated (20%). No other anomaly was reported in 3 cases of 22q11.2 deletion. Apart from congenital heart defects, the commonest structural anomalies present were cleft soft palate (4 cases) and absent thymus (3 cases).

Table 1: Types of heart defects associated with 22q11.2 deletion

Heart Defect	**Frequency**	**Number resulting in live birth**
Ventricular septal defect	15	13
Tetralogy of fallot	6	5
Interruption of aorta	6	6
Truncus arteriosus	4	1
Atrial septal defect	4	4
Double outlet right ventricle	3	3
Pulmonary valve atresia	3	3
Pulmonary artery atresia	3	2
Major aortopulmonary collateral arteries	3	2
Patent foramen ovale (in term babies)	3	3
Pulmonary valve stenosis	2	2
Overiding aorta	2	2
Persistent right aortic arch	2	2
Pulmonary artery stenosis	2	2
Enlarged right atrium and / or ventricle	2	2
Patent ductus arteriosus (in term babies)	2	2
Bicuspid aortic valve	2	2
Aortic valve stenosis	1	1
Subaortic stenosis	1	1
Hypoplasia of aorta	1	1
Dysplastic pulmonary valve	1	1
Persistent left superior vena cava	1	1

Table 2: Comparison of q2211.2 deletion with / without heart defects

	CDH without heart anomalies	CDH with heart anomalies
Antenatal detection of q2211.2	20% (2/10)	23% (7/30)
Survival to 3 years (1998-2008 live births)	75% (6/8)	78% (21/27)

Conclusion: 22q11.2 deletion commonly presents with congenital heart anomalies. Karyotyping for the deletion should be offered to babies presenting with heart disease. Antenatal detection of the deletion and survival is similar for all cases irrespective of the presence of congenital heart anomalies.