

Extra-cardiac malformations, chromosomal abnormalities and clinical syndromes in neonates with congenital heart disease

*Panagiotopoulou O., Karatza A., Mantagou L., Tzifas S., Sinopidis X., Dimitriou G., Mantagos S.
Department of Paediatrics, University of Patras Medical School, Rio, Patras, Greece*

Introduction: Congenital heart disease (CHD) is frequently associated with extra-cardiac malformations, chromosomal abnormalities and recognised clinical syndromes. The management of the heart lesion may be influenced by the treatment needed for the associated anomalies and vice versa. A multidisciplinary approach may be warranted and counselling of the family regarding long-term outcome should be modified accordingly. The aim of this study is to assess the incidence of extra-cardiac malformations, chromosomal abnormalities and clinical syndromes in neonates with congenital heart disease.

Methods: Data on all neonates referred for echocardiography to our institution were collected prospectively. Infants with abnormal echocardiograms were followed up in clinic and were managed appropriately. Information regarding the presence of chromosome abnormalities, recognised syndromes and extra-cardiac defects was recorded. The classification of birth defects was performed using the EUROCAT (European Surveillance of congenital anomalies) Registry Guide 1.3.

Results: Over a period of 5.5 years (6/2006-12/2011) 866 neonates (aged 1-28 days) were referred for echocardiography, of which 305 had CHD (35.2%). Two hundred fifty nine patients (84.9%) had isolated CHD. Nineteen (6.2%) had chromosome abnormalities, 5/305 (1.6%) had clinically recognised syndromes and 22/305 (7.2%) had extra-cardiac anomalies not associated with genetic abnormalities or clinical syndromes. Among those infants the indication for echocardiography was the identification of the extra-cardiac lesion or suspected syndrome in 21/46 (45.6%). The most common genetic or clinical syndrome associated with CHD was Trisomy 21 (13/24, 51.2%). In this group the most prevalent cardiac anomaly was atrioventricular septal defect (7/24, 21.2%), whereas in infants with extra-cardiac anomalies not associated with genetic or clinical syndromes the most common diagnosis was interventricular septal defect (11/22, 50%). Anomalies of the gastrointestinal system were the most prevalent in the latter group (7/22, 31.8%).

Conclusion: About 15% of neonates with CHD have extra-cardiac anomalies, chromosomal abnormalities or recognised syndromes. Neonatologists and paediatric surgeons have a low threshold for echocardiographic referral of patients with extra-cardiac anomalies or syndromes associated with increased risk of CHD.