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Clinical, Cytogenetic and Molecular studies in Egyptian Children with Congenital Heart Disease (CHD)

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The cardiovascular anomalies responsible for congenital heart disease (CHD) are common birth defects and are a leading cause of infant mortality, morbidity. CHD is a primary feature of many genetic syndromes, but genetic causes of non-syndromic CHD are increasingly being recognized. CHD affects at least one in 100 live births. It ranges widely in severity, from tiny holes between heart chambers that close naturally, to life-threatening abnormal structures such as hypoplastic left-heart syndrome. Recently identified disease genes for syndromes associated with congenital heart disease are also increasing. These gene discoveries are being rapidly translated into meaningful genetic testing, improving the diagnosis and prognostication for CHD. To date, very few studies were performed in Egypt for studying the genetic basis of CHD. This study included 500 cases with CHD out of total 8000 cases referred as genetic disorder to the Clinical Genetic Department of the NRC during a period of 6 years (2004-2010). It aims to identify at risk families and types of syndromes with CHD, identify types of chromosomal anomalies causing these diseases & study the spectrum of mutations in known candidate genes. Chromosomal analysis was performed on 200 cases. Molecular studies were carried out for 189 cases. Mutation detection was done using PCR/ SSCP and DNA sequencing. Associated malformations as known syndromes included Russell Silver, Jacobsen, Noonan, Costello, Apert, Kamptomelic dysplasia, McKusik Kaufman, Rubinstien-Taybi, William, Holt-Oram & Roberts. Chromosomal anomalies reported are trisomy 21, trisomy 13, trisomy 18, 46, XY, t (14;18), 46, XYt (8;9), 47, XX, + mar ; 46, XX del 11q ,46, XX, dup13q, 47, XXX, 45, X, 47XXY, 46, XX, inv(18). DNA sequencing of NKX2.5 gene revealed two different mutations in two unrelated patients. Sequencing of GATA4 gene in patients with TOF has shown polymorphism in exon 6 at nucleotide 53423 (A-G) in 5 patients out of 19 cases. The genetic investigation results of Nkx2.5 gene concluded the presence of missense mutation in one out of 19 TOF cases (5.3%), two missense mutation in two out of 35 ASD cases (5.7%) and one silent mutation in one out of 35 ASD cases (2.9%). Results of GATA4 gene concluded the presence of two silent mutations in two out of 19 TOF cases (10.5%), and three SNPs in 9 out of 19 TOF cases (47.4%). Results of 22q11.2 critical DiGeorge region (CDGR) concluded the presence of microdeletion in 6 out of 18 DiGeorge/ VCF syndrome cases (33.3%). Results of PTPN11 gene concluded the presence of Polymorphism -21 (C→T) in one out of 16 NS cases (6.3%).