Conotruncal heart malformations

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Conotruncal heart malformations. ORPHA:2445

Conotruncal heart malformations are a group of congenital cardiac outflow tract anomalies that include such defects as tetralogy of Fallot, pulmonary atresia with ventricular septal defect, double-outlet right ventricle (DORV), double-outlet left ventricle, truncus arteriosus and transposition of the great arteries (TGA) (see these terms), among others. This group of defects is frequently found in patients with 22q11.2 deletion syndrome (see this term). A deletion of chromosome 22q11.2 has equally been associated in a subset of patients with various types of isolated non-syndromic conotruncal heart malformations (with the exception of DORV and TGA where this is very uncommon).