Opis choroby *

Definicja

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), among others. This group of defects is frequently found in patients with 22q11.2 deletion syndrome . 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).

Dane

Klasyfikacja
Kategoria

Kod ORPHA
2445

Kod OMIM
217095

Kod ICD10
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Kod ICD11
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*Źródło

orphanet