NCCU CLINICAL GUIDELINES
SECTION: 14

NEONATAL CARDIAC CONDITIONS: MEDICAL AND SURGICAL MANAGEMENT

Section 14 Neonatal cardiac conditions
Congenital heart disease: Syndromes and cardiac defects
Date created: Oct 2014
Date revised:
Review date: Oct 2017

Neonatology Clinical Guidelines
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SYNDROMES ASSOCIATED WITH CONGENITAL CARDIAC DEFECTS

	ASSOCIATED CARDIAC ANOMALIES
TRISOMY 13 (PATAU SYNDROME) Low birth weight, microcephaly with sloping forehead, broad flat nose, scalp defects, CNS malformations, eye malformations, cleft lip/ palate, polydactyly, low-set ears, visceral and genital anomalies. 80% have CHD.	VSD / PDA / dextrocardia
TRISOMY 18 (EDWARD SYNDROME) Low birth weight, microcephaly, micrognathia, short palpebral fissues, small mouth, rocker-bottom feet, closed fist with overlapping fingers. 90% have CHD.	VSD/ PDA/ PS
TRISOMY 21 (DOWN SYNDROME) Mental impairment, hypotonia, epicanthic folds, flat nasal bridge, small ears, transverse palmer crease. 40-50% have CHD.	VSD/ ASD/ AVSD/ PDA/ TOF Pulmonary vascular disease
TURNER SYNDROME (46 XO) Mental impairment, ambiguous genitalia, short stature, webbed neck, broad chest with widely spaced nipples. Oedema of hands and feet in neonates. 35% have CHD.	CoA (20%) Bicuspid aortic valve (35%)/ AS/ ASD. Less commonly: anomalous pulmonary valve connection / HLHS / mitral valve abnormalities
22Q11 DELETION (DIGEORGE/ VELO-CARDIO-FACIAL/ CATCH 22) Hypoplastic or absent thymus causing deficient cell-mediated immunity and hypoplastic or absent parathyroids causing hypocalcaemia (50%). Bulbous nose, anteverted palpebral fissures, small and/ or low-set ears, cleft palate (50%), small stature. Learning difficulties (90%). Renal anomalies (37%). 40% have CHD. 3% of children with CHD have this deletion.	Conotruncal malformations: Truncus arteriosus/ IAA/ CoA/ DORV/ PA + VSD/ TOF
CRIT-DU-CHAT SYNDROME 5q deletion. Striking craniofacial dysmorphia with downward slanting palebral fissures and microcephaly, 'cat-like' cry, low birth weight, failure to thrive, developmental delay. 25% have CHD.	VSD/ ASD/ bicuspid aortic valve/ TOF/ PDA

WILLIAM'S SYNDROME May be a 7q deletion or AD 6;7 translocation. Connective tissue disorder with deletion of elastin gene. Elfin facies (upturned nose, flat nasal bridge, long philtrum, flat malar area, wide mouth, full lips, widely spaced teeth, periorbital fullness), mental impairment, 'cocktail' personality. ?Hypercalcaemia of infancy.	Supravalvular AS (66%)/ supravalvular PS/ PA stenosis/ VSD/ PDA. Diffuse arterial wall thickening involving coronary arteries and RAS. Systemic hypertension.
WOLF-HIRSCHHORN SYNDROME 4p deletion. Facial dysmorphia, severe growth and mental impairment, microcephaly, hypotonia, weak suck. 50% have CHD.	VSD/ ASD/ PDA/ PS
SINGLE GENE DEFECTS	
ALAGILLE SYNDROME (ARTERIOHEPATIC DYSPLASIA) AD inheritance. Peculiar facies (95%) (deep-set eyes, broad forehead, long straight nose with flattened tip, prominent chin, small low-set malformed ears). Paucity of intrahepatic bile ducts with cholestasis (91%). Hypercholesterolaemia. Butterfly-like vertebral arch defects (87%). Growth retardation (50%). Mild mental impairment (16%). 85% have CHD.	Peripheral PA stenosis +/- complex CV abnormalities
CARPENTER SYNDROME (ACROCEPHALOSYNDACTYLY TYPE II) AR inheritence. Brachycephaly with variable craniosynostosis and mild facial hypoplasia. Polydactyly and severe syndactyly (mitten hand). 50% have CHD.	PDA/ VSD/ PS/ TGA
CORNELIA DE LANGE SYNDROME Synophrys, hirsuitism, prenatal growth retardation, microcephaly, anteverted nares, downturned mouth, mental impairment. 30% have CHD.	VSD
EHLERS-DANLOS SYNDROME TYPE IV Defect in type III collagen. AD inheritance. Hyperextensibility of skin and hypermobility of joints.	MV prolapse common, aortic root dilatation. Spontaneous rupture of large and medium calibre arteries.
ELLIS-VAN CREVELD SYNDROME (CHONDROECTODERMAL DYSPLASIA) AR inheritance. Short stature, short distal extremities, narrow thorax with short ribs, polydactyly, nail hypoplasia, neonatal teeth. 50% have CHD.	ASD/ single atrium
GLYCOGEN STORAGE DISEASE TYPE II (POMPE DISEASE) Large tongue, hypotonia, weak muscles, respiratory distress, cardiomegaly. AR inheritance.	Cardiomyopathy – severe ventricular hypertrophy on echo

HOLT-ORAM SYNDROME (CARDIO-LIMB SYNDROME) Deletion in HOS1 gene. AD inheritance. Defects or absence of thumb or radius.	ASD/ VSD/ AVSD/ TOF/ conduction abnormalities
LEOPARD SYNDROME (MULTIPLE LENTIGINES SYNDROME) **L**entiginous skin lesion **E**CG abnormalities **O**cular hypertelorism **P**ulmonary stenosis **A**bnormal genitalia **R**etarded growth **D**eafness	PS/ HOCM/ long PR interval
MARFAN SYNDROME Connective tissue defect resulting from mutation in fibrillin gene. AD inheritance with clinical variability. Kyphoscoliosis, pectus carinatum, arachnoidactyly, high arched palate, hypermobility, lens dislocation.	Progressive aortic root dilatation leading to aortic dissection. MV prolapse.
NOONAN'S SYNDROME Short stature, hypertelorism, low-set ears, ptosis. AD inheritance/ sporadic.	PS (39%)/ hypertrophic cardiomyopathy (10%)/ ASD (8%)/ TOF (4%)/ CoA (9%)/ MV anomalies (6%)
RUBENSTEIN-TAYBI SYNDROME Broad thumbs/ toes, hypoplastic maxilla with narrow palate, beaked nose, short stature, mental impairment. AD inheritance. 25% have CHD.	PDA/ VSD/ ASD
SMITH-LEMLI-OPITZ SYNDROME Microcephaly, broad nasal tip with anteverted nostrils, ptosis of eyelids, syndactyly of 2nd and 3rd toes, hypotonia, short stature, mental impairment. Genitourinary abnormalities. AR inheritance.	VSD/ PDA