



CLINICAL PRACTICE GUIDELINE

Guideline coverage includes NICU KEMH, NICU PMH and NETS WA


Cardiac: Syndromes Associated with Congenital Cardiac Defects

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Syndrome	Associated Cardiac Anomalies
<p>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.</p>	VSD / PDA / dextrocardia
<p>Trisomy 18 (Edward Syndrome) Low birth weight, microcephaly, micrognathia, short palpebral fissures, small mouth, rocker-bottom feet, closed fist with overlapping fingers. 90% have CHD.</p>	VSD / PDA / PS
<p>Trisomy 21 (Down Syndrome) Mental impairment, hypotonia, epicanthic folds, flat nasal bridge, small ears, transverse palmer crease. 40-50% have CHD.</p>	VSD / ASD / AVSD / PDA / TOF Pulmonary vascular disease
<p>Turner Syndrome (46XO) Mental impairment, ambiguous genitalia, short stature, webbed neck, broad chest with widely spaced nipples. Oedema of hands and feet in neonates. 35% have CHD.</p>	CoA (20%) Bicuspid aortic valve (35%) / AS / ASD Less commonly: anomalous pulmonary valve connection / HLHS / mitral valve abnormalities
<p>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.</p>	Conotruncal malformations: Truncus arteriosus / IAA / CoA / DORV / PA + VSD / TOF

<p>Crit-du-Chat Syndrome 5q deletion. Striking craniofacial dysmorphism with downward slanting palpebral fissures and microcephaly, 'cat-like' cry, low birth weight, failure to thrive, developmental delay. 25% have CHD.</p>	<p>VSD / ASD / Bicuspid aortic valve / TOF / PDA</p>
<p>William's Syndrome Maybe 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.</p>	<p>Supravalvular AS (66%) / supravalvular PS / PA stenosis / VSD / PDA. Diffuse arterial wall thickening involving coronary arteries and RAS. Systemic hypertension.</p>
<p>Wolf-Hirschhorn Syndrome 4p deletion. Facial dysmorphism, severe growth and mental impairment, microcephaly, hypotonia, weak suck. 50% have CHD.</p>	<p>VSD / ASD / PDA / PS</p>
<p>Single Gene Defects</p>	<p>Associated Cardiac Anomalies</p>
<p>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.</p>	<p>Peripheral PA stenosis +/- complex CV abnormalities</p>
<p>Carpenter Syndrome (Acrocephalosyndactyly Type II) AR inheritance. Brachycephaly with variable craniosynostosis and mild facial hypoplasia. Polydactyly and severe syndactyly (mitten hand). 50% have CHD.</p>	<p>PDA / VSD / PS / TGA</p>
<p>Cornelia de Lange Syndrome Synophrys, hirsutism, prenatal growth retardation, microcephaly, anteverted nares, downturned mouth, mental impairment. 30% have CHD.</p>	<p>VSD</p>
<p>Ehlers-Danlos Syndrome Type IV Defect in type III collagen. AD inheritance. Hyperextensibility of skin and hypermobility of joints.</p>	<p>MV prolapse common, aortic root dilation. Spontaneous rupture of large and medium calibre arteries.</p>

<p>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.</p>	<p>ASD / single atrium</p>
<p>Glycogen Storage Disease Type II (Pompe Disease) Large tongue, hypotonia, weak muscles, respiratory distress, cardiomegaly. AR inheritance.</p>	<p>Cardiomyopathy - severe ventricular hypertrophy on echo</p>
<p>Holt-Oram Syndrome (Cardio-limb Syndrome) Deletion in HOS1 gene. AD inheritance. Defects or absence of thumb or radius.</p>	<p>ASD / VSD / AVSD / TOF / conduction abnormalities</p>
<p>LEOPARD Syndrome (Multiple Lentiginos Syndrome) Lentiginous skin lesion, ECG abnormalities, Ocular hypertelorism, Pulmonary stenosis, Abnormal genitalia, Retarded growth, Deafness</p>	<p>PS / HOcm / long PR interval</p>
<p>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.</p>	<p>Progressive aortic root dilatation leading to aortic dissection. MV prolapse.</p>
<p>Noonan's Syndrome Short stature, hypertelorism, low-set ears, ptosis. AD inheritance / sporadic.</p>	<p>PS (39%) / hypertrophic cardiomyopathy (10%) / ASD (8%) / TOF (4%) / CoA (9%) / MV anomalies (6%)</p>
<p>Rubenstein-Taybi Syndrome Broad thumbs / toes, hypoplastic maxilla with narrow palate, beaked nose, short stature, mental impairment. AD inheritance. 25% have CHD.</p>	<p>PDA / VSD / ASD</p>
<p>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.</p>	<p>VSD / PDA</p>

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