GUIDELINE

Cardiac: Syndromes Associated with Congenital Cardiac Defects

Scope (Staff):	Nursing and Medical Staff
Scope (Area):	NICU KEMH, NICU PCH, NETS WA

Child Safe Organisation Statement of Commitment

CAHS commits to being a child safe organisation by applying the National Principles for Child Safe Organisations. This is a commitment to a strong culture supported by robust policies and procedures to reduce the likelihood of harm to children and young people.

This document should be read in conjunction with this disclaimer

Aim

To provide guidance on the clinical process of investigation and referral to cardiology of infants at increased risk of a congenital heart disease, in association with genetic syndromes.

Risk

Failure to monitor and detect abnormalities can result in delay treatment or result in poor health outcomes. Delayed diagnosis can be associated with increased mortality and morbidity from multi-organ damage.

Background

- One third of cases of congenital heart disease occur in association with chromosomal anomalies and genetic syndromes.
- Infants with complex CHD (including those who need surgical intervention in the first year of life and those with other co-morbid physical conditions) are at increased risk of neurodevelopmental impairment and disability.
- The risk and severity of neurological impairment increase with greater CHD complexity and the presence of a genetic disorder or syndrome.

Management

- All infants with a suspected or confirmed genetic anomaly with known increased association of congenital heart disease should undergo a comprehensive clinical assessment of cardiovascular examination (auscultation, pulses, BP, pulse oximetry) and supplementary investigations, where indicated (CXR, EEG) and specialist referral.
- Where a cardiology opinion is requested, for infants in whom there is a strong suspicion of congenital or acquired heart disease, this should be provided by a paediatric cardiologist, to ensure that appropriate advice is provided to the infant's family and management is optimised. As an example, the Paediatric and Congenital Council recommends that all infants with Trisomy 21 be seen before two months of age by a physician with experience in the examination of the cardiovascular system of the newborn and echocardiography performed by a paediatric cardiologist or paediatric sonographer (under the guidance of a paediatric cardiologist) as per the Standards of Practice for Paediatric Echocardiography (CSANZ/ASUM).

Genetics **Chromosomal Aneuploidies**	**Associated Cardiac Anomalies**	**Reported CHD Prevalence**
Trisomy 13 (Patau Syndrome) Low birth weight, microcephaly with sloping forehead, broad flat nose, scalp defects, midline anomalies (holoprosencephaly), colobomas, cleft lip/palate, polydactyly, low-set ears, genital anomalies.	Dextrocardia/ VSD / PDA	80%
Trisomy 18 (Edward Syndrome) Low birth weight, severe microcephaly, hypotonia, micrognathia, short palpebral fissures, small mouth & jaw, redundant skin folds, rocker-bottom feet, clenched fist with overlapping fingers.	VSD / PDA / PS	90%
Trisomy 21 (Down Syndrome) Neonatal hypotonia, characteristic facies of epicanthic folds, upward-slanting palpebral fissures, flat nasal bridge, small ears, transverse palmer crease.	VSD / ASD / AVSD / PDA / TOF PPHN	40-50%
Turner Syndrome (46XO) Dorsal lymphoedema of the hands and feet, redundant skin folds, webbed neck, broad chest with widely spaced nipples, nail hypoplasia.	Coarctation of the Aorta with bicuspid aortic valve / AS /ASD	30%

Chromosomal Deletion Syndromes		
Crit-du-Chat Syndrome (5p- deletion) Low birth weight, microcephaly, craniofacial dysmorphia with downward slanting palpebral fissures, low set ears, 'cat-like' cry, syndactyly, failure to thrive.	VSD / ASD / Bicuspid aortic valve / TOF / PDA	25%
Wolf-Hirschhorn Syndrome (4p- deletion). Microcephaly, hypotonia, weak suck. Facial dysmorphia, colobomas, cleft palate, hypospadias, cryptorchidism	VSD / ASD / PDA / PS	50%
Microdeletions		
Alagille Syndrome (20p.12) AD inheritance. Peculiar facies with deep-set eyes, broad forehead, long straight nose with flattened tip, prominent chin, small low-set malformed ears. Cholestasis/ bile duct paucity, butterfly vertebra.	Peripheral PA stenosis +/- complex CV abnormalities	85%
22q11.2 Deletion Syndrome Hypoplastic/absent thymus and parathyroid glands (immune dysfunction/ hypocalcaemia/ hypoparathyroidism). Bulbous nose, anteverted palpebral fissures, small low-set ears, cleft palate.	Conotruncal malformations: Truncus arteriosus / IAA / CoA / DORV / PA + VSD / TOF	75-85%
Jacobsen Syndrome (11q-) IUGR, hypertelorism, broad nasal bridge, thin lip, thrombocytopenia.	HLHS / AS / VSD/ CoA	50%
Rubenstein-Taybi Syndrome (16p13-) Broad thumbs, large toes, hypoplastic maxilla with narrow palate, beaked nose, short stature.	PDA / VSD / ASD	25%
William's Syndrome (7q11.23 deletion) Connective tissue anomalies, elfin facies (upturned nose, flat nasal bridge, long philtrum, flat malar area, wide mouth, full lips, periorbital fullness), transient hypercalcaemia.	Supravalvular Aortic or Pulmonary Stenosis/ VSD / PDA. Diffuse arterial wall thickening involving coronary	80%

	arteries. Systemic hypertension.	
Syndromes		
Carpenter Syndrome (Acrocephalosyndactyly Type II) AR inheritance, abnormal bone fusion with craniosynostosis, abnormal facial features, polydactyly and syndactyly, hearing, and visual impairment.	VSD / PS / TGA	50%
CHARGE Multisystem anomalies, coloboma, choanal atresia, cardiac/ear/renal/GU anomalies, IUGR.	TOF/ PDA/ DORV/ AVSD	75-85%
Cornelia de Lange Syndrome IUGR, microcephaly, synophrys, hirsutism, anteverted nares, downturned mouth, micrognathia, toe syndactyly.	ASD/ VSD	30%
Costello Syndrome (11p15.5) IUGR, poor feeding, broad face with bitemporal narrowing, redundant skin.	PS/ ASD/ VSD	45%
Ellis Van Creveld Syndrome (Chondroectodermal Dysplasia) AR inheritance affecting bone growth. Short stature, short limbs, narrow chest, short ribs, polydactyly, nail hypoplasia, neonatal teeth.	AVSD / single atrium	50%
Glycogen Storage Disease Type II (Pompe Disease) AR inheritance affecting muscle, heart, liver and nerves. Large tongue, hypotonia, weak muscles, respiratory distress, poor feeding, cardiomegaly.	Cardiomyopathy, severe ventricular hypertrophy	80-90%
Holt-Oram Syndrome (Cardio-limb Syndrome) AD inheritance. Upper limb defects, particularly radial/ thumbs and heart.	ASD / VSD / AVSD / TOF / conduction abnormalities	70-80%
LEOPARD Syndrome (Multiple Lentigines Syndrome) Abnormalities of skin, heart, inner ears and genitalia. (**L**entiginous skin lesion, **E**CG abnormalities, **O**cular hypertelorism, **P**ulmonary stenosis, **A**bnormal genitalia, **R**etarded growth, **D**eafness)	Hypertrophic cardiomyopathy/ PS / long PR interval	80%

Noonan's Syndrome Short stature, hypertelorism, low-set ears, ptosis. AD inheritance / sporadic.	PS / hypertrophic cardiomyopathy / ASD / TOF / CoA / MV anomalies	75%

Related CAHS internal policies, procedures and guidelines

Neonatology Guidelines

- Admission to NICU KEMH and PCH
- Cardiac Arrest and Arrhythmias in NICU: Treatment Algorithms
- Cardiac Arrhythmias
- Cardiac Cardioversion and Defibrillation
- Cardiac: Blalock-Taussig Shunt (BT Shunt) Management Following Insertion
- Cardiac: Care Post Cardiac Catheterisation
- Cardiac: Coarctation of the Aorta (CoA) and Interrupted Aortic Arch (IAA)
- Cardiac: Coarctation of the Aorta (CoA) Management Following Surgical Repair
- Cardiac: Complications Management Following Surgery
- Cardiac: Management of Balloon Atrial Septostomy for Congenital Heart Defects in the NICU
- Cardiac: Neonatal Circulation Changes / Unbalanced Circulation
- Cardiac: Post-Operative Handover
- Cardiac: Routine Post-Operative Care

References and related external legislation, policies, and guidelines

- Paediatric Cardiology - Standards of Practice Statement (csanz.edu.au) The Cardiac Society of Australia and New Zealand accessed 09/08/2023
- Australian Institute of Health and Welfare 2019. Congenital heart disease in Australia. Cat. no. CDK 14. Canberra: AIHW. Congenital heart disease in Australia (Full publication;[15October2019]edition)(AIHW)
- Yasuhara J, Garg V. Genetics of congenital heart disease: a narrative review of recent advances and clinical implications. Transl Pediatr. 2021 Sep;10(9):2366-2386. doi: 10.21037/tp-21-297. PMID: 34733677; PMCID: PMC8506053 Genetics of congenital heart disease: a narrative review of recent advances and clinical implications - PMC (nih.gov)

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