



Common Genetic Syndromes and Associated Heart Defects

A Pediatric Cardiology Reference Guide

Recognizing the association between genetic syndromes and congenital heart defects (CHDs) helps with early diagnosis, appropriate screening, and multidisciplinary care.



Genetic Syndrome	Common Associated Cardiac Abnormalities	Key Features / Notes	Examples of Key Heart Defects
Down syndrome (Trisomy 21)	Atrioventricular septal defect (AVSD), VSD, ASD, PDA, tetralogy of Fallot (TOF)	AVSD is characteristic.	Atrioventricular Septal Defect (common in Down syndrome)
Turner syndrome (45,X)	Bicuspid aortic valve, coarctation of the aorta, aortic dilation, partial anomalous pulmonary venous return	Left-sided obstructive lesions; monitor aorta lifelong.	Coarctation of the Aorta (common in Turner syndrome)
DiGeorge syndrome (22q11.2 deletion)	TOF, truncus arteriosus, interrupted aortic arch (especially type B), VSD, right aortic arch	Conotruncal defects are typical.	Tetralogy of Fallot (common in 22q11.2 deletion)
Williams syndrome (7q11.23 deletion)	Supravalvular aortic stenosis, peripheral pulmonary artery stenosis, other arterial stenoses	Diffuse arteriopathy; anesthesia considerations.	Supravalvular Aortic Stenosis (common in Williams syndrome)
Noonan syndrome	Pulmonary valve stenosis (often dysplastic), hypertrophic cardiomyopathy (HCM), ASD	Variable expression; serial follow-up for HCM.	Hypertrophic Cardiomyopathy (common in Noonan syndrome)
Marfan syndrome	Aortic root dilation, aortic aneurysm/dissection, mitral valve prolapse, mitral regurgitation	Aortopathy; lifelong monitoring.	
Loeys-Dietz syndrome	Aortic root aneurysm, arterial tortuosity, aneurysms and dissections throughout the arterial tree	Aggressive vascular disease; early surveillance.	
Ehlers-Danlos syndrome (vascular type)	Arterial aneurysm, dissection, rupture	Vascular fragility; spontaneous dissections.	
Holt-Oram syndrome	Secundum ASD, VSD, conduction abnormalities, atrioventricular block	Associated upper limb anomalies.	
Alagille syndrome	Peripheral pulmonary artery stenosis, TOF, other pulmonary arterial abnormalities	Cholestasis, characteristic facies.	
CHARGE syndrome	TOF, AVSD, VSD, aortic arch anomalies, complex CHDs	Multiple system involvement.	
Trisomy 18 (Edwards syndrome)	VSD, PDA, ASD, polyvalvular abnormalities	Often multiple anomalies.	
Trisomy 13 (Patau syndrome)	VSD, ASD, PDA, complex CHDs	Often severe, multisystem anomalies.	
Kabuki syndrome	Coarctation of the aorta, ASD, VSD, other left-sided obstructive lesions	Distinctive facial features.	
Rubinstein-Taybi syndrome	ASD, VSD, PDA and other congenital heart defects	Broad phenotypic variability.	
Ellis-van Creveld syndrome	Common atrium, AVSD, ASD	Skeletal dysplasia, short stature.	
Smith-Lemli-Opitz syndrome	AVSD, ASD, VSD and other congenital heart defects	Disorders of cholesterol synthesis.	
Tuberous sclerosis complex	Cardiac rhabdomyomas, possible arrhythmias or ventricular outflow obstruction	Often detected prenatally or in infancy.	
Pompe disease (infantile-onset)	Marked hypertrophic cardiomyopathy, conduction abnormalities	Later-onset forms may have milder cardiac disease.	
Friedreich ataxia	Hypertrophic cardiomyopathy, myocardial fibrosis, arrhythmias	Progressive cardiomyopathy.	

Clinical Evaluation and Screening

- Detailed family history
- Physical examination (murmurs, pulses, blood pressure)
- ECG
- Echocardiography
- Consider genetic consultation and targeted testing

Long-Term Management

- Individualized follow-up based on syndrome and cardiac findings
- Serial echocardiograms, ECGs
- Ambulatory rhythm monitoring when indicated
- Cardiac MRI or CT for aortic or vascular assessment
- Multidisciplinary care

Clinical Pearls

- Screen for CHDs even if no murmur in at-risk syndromes.
- Monitor aortic size in Marfan, Loeys-Dietz, Turner, and other aortopathy syndromes.
- Patients with Down syndrome and large left-to-right shunts may develop pulmonary hypertension early.
- Consider anesthesia risk in Williams syndrome.
- Endocarditis prophylaxis is indicated only for specific high-risk conditions.

Remember

- These are characteristic associations, not present in every patient.
- Some conditions primarily cause acquired or progressive cardiovascular disease rather than structural congenital defects.
- Early recognition improves outcomes through timely intervention and coordinated care.



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+1 (555) 123-4567
pedsheart@yourhospital.org
www.yourhospital.org/pediatric-cardiology

