# [Congenital Heart Disease: Prenatal Diagnosis and Genetic Associations.](https://www.ncbi.nlm.nih.gov/pubmed/31418452)

Hopkins MK, Dugoff L, Kuller JA.

Obstet Gynecol Surv. 2019 Aug;74(8):497-503. doi: 10.1097/OGX.0000000000000702.

PMID: 31418452

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**Take Home Points:**

* If congenital heart disease (CHD) is suspected on prenatal testing, parents should be referred for genetic counselling and/or genetic testing.
* There are several maternal risk factors for fetal CHD including advanced maternal age, diabetes mellitus, obesity and exposures (ex: alcohol and cocaine).
* Common genetic conditions associated with a prenatal diagnosis of CHD include chromosomal abnormalities and single-gene defect disorders.



**Commentary from Dr. Charlotte Van Dorn (Rochester, MN), section editor of Pediatric Cardiology Journal Watch:** Congenital heart disease (CHD) remains the most common birth defect (approximately 4-50 per 1000 live births) and is associated with several genetic syndromes. The early detection of CHD can allow for appropriate perinatal referrals and can improve survival in infants; early detection can also provide information regarding post-natal prognosis.

Risk factors for CHD include a family history of CHD, coexisting maternal disease (ex: diabetes mellitus), teratogen exposure (ex: lithium), advanced maternal age, monochorionic twinning, maternal obesity, maternal alcohol and drug use (ex: cocaine) and in vitro fertilization. Fetal echocardiography should be obtained if there is concern for an abnormal cardiac screening ultrasound (US) and/or predisposition to genetic CHD. The detection of other structural abnormalities by prenatal US increases the risk of genetic syndromes associated with CHD.

Chromosomal disorders (trisomy 13, 18 and 21) make up 8-10% of CHD. These conditions, especially trisomy 21, can be associated with advanced maternal age. Approximately 50% of infants with trisomy 21 will have CHD, while up to 85% of infants with trisomy 13 and up to 94% of infants with trisomy 18 will have CHD. More common defects include atrial and ventricular septal defects as well as patent ductus arteriosus. One third of patients with trisomy 13 and up to 24% of patients with trisomy 18 will have complex CHD.

Turner syndrome, involving complete or partial absence of one X chromosome, has a CHD incidence of 23-50%. This most commonly involves left-sided obstructive lesions such as bicuspid aortic valve (12-18%) and coarctation of the aorta (7-18%). CHD is also common in DiGeorge syndrome (22q11.2 deletion syndrome) occurring in up to 81% of patients. Infants born with conotruncal defects have a 50% likelihood of having DiGeorge Syndrome.

Single-gene defect disorders account for 3-5% of CHD and can often be associated with noncardiac malformations. Common single-gene defect disorders include *Noonan Syndrome*, *Alagille Syndrome*, and *Holt-Oram Syndrome*.

Perinatal genetic testing options include karyotype, chromosomal microarray (CMA) and amniocentesis. These tests do not detect every genetic disease or syndrome and for single-gene defects, specific gene panels and/or whole exome sequencing may be required. Genetic testing should not be performed without pretest genetic counselling.



