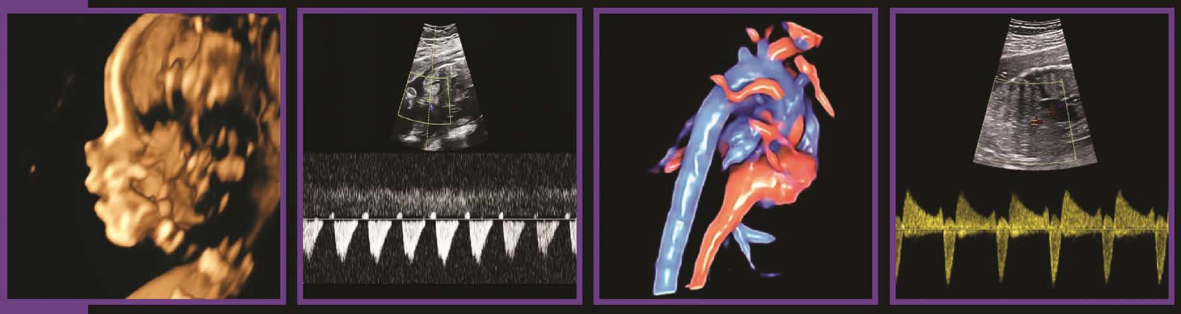
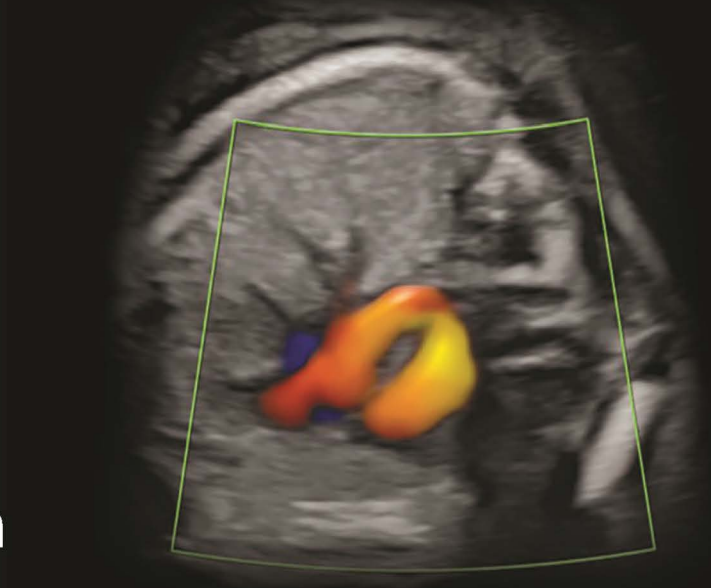
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Fetal Echocardiography



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2nd Edition

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Chapter

10

Genetics and Congenital Heart Disease

Pradeep S, SJ Patil, Meenakshi Bhat

APPROACH: TO EVALUATE FOR EXTRACARDIAC MALFORMATIONS AND CHROMOSOME ANOMALIES AFTER DIAGNOSIS OF FETAL CONGENITAL HEART DISEASE

Pradeep S

Abstract

- The scope of the chapter is to introduce to the readers about the association of findings in fetuses with cardiac anomalies.
- Cardiac anomalies can occur in isolation or in association with other anomalies. The associated anomalies may be chromosomal abnormalities or physical abnormalities or physical abnormalities constituting a genetic syndrome.
- When associated with chromosomal abnormalities the cardiac anomaly may be isolated or may be associated with other physical abnormalities.
- Among various genetic syndromes, commonest the few are described in this chapter.
- Extracardiac abnormalities in association with cardiac anomalies can involve gastrointestinal system, genitourinary system, respiratory system, musculoskeletal system, craniofacial, and central nervous system (CNS).
- Among various chromosomal abnormalities, trisomy 21, trisomy 18, trisomy 13, trisomy 22, 22Q deletion, Turner's 45,X0, and triploidy are common associations in fetuses with cardiac anomalies and are described in the text.
- When cardiac anomalies are encountered, attempt should be made to evaluate the entire fetus to detect all other associated abnormalities in extracardiac systems. Invasive testing by chorionic villus sampling (CVS), amniocentesis, or cordocentesis has to be done to exclude associated aneuploidy **(Tables 1 and 2)**. Noninvasive prenatal cell-free DNA analysis may be selectively used in certain appropriate circumstances for aneuploidy and single-gene disorder detections.
- History of known genetic disorders in family or siblings can help in exclusion of the same in the fetus.
- Prognosis and counseling can be complete with knowledge of presence of associated extracardiac anomalies and chromosomal/genetic abnormalities.

Keywords: Chromosomal anomalies, congenital heart disease, extracardiac anomalies

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INTRODUCTION

Cardiac anomalies are frequently encountered during antenatal sonography. Advanced equipment, high-frequency transducers, transvaginal ultrasound, fetal echo sonography settings, volume cardiac acquisition, sectional planes and postprocessing better training, and experienced hands have made detection of cardiac anomalies a reality in the first trimester from 14 weeks onward.

With such capabilities and early diagnosis of cardiac anomalies, it is very important to complete the analysis by determining the presence or absence of extracardiac associated anomalies. This can be achieved by performing a detailed targeted imaging for fetal anomalies (TIFFA) in a systematic manner, systemwise with documentation of a minimum of 40 pictures and a minimum of 15 measurements called as the extended biometry. Extended biometry includes the following measurements—biparietal diameter (BPD), head circumference, occipito frontal diameter, interorbital distance, binocular diameter, nasal bone (NB) length, ear length, cerebral lateral ventricle, transcerebellar diameter, cisterna magna, intracranial lucency, facial angle, prenasal thickness, nuchal lucency, nuchal fold thickness, thoracic circumference, kidney bipolar lengths, femur length (FL), tibia length, foot length, humerus length, radius length. From the above measurements, cephalic index, FL/foot ratio, BPD/FL ratio, abdominal circumference (AC)/FL ratio, etc. can be calculated.

In the first trimester, aneuploidy markers are increased nuchal lucency thickness, NB absence, increased prenasal thickness, abnormal intracranial lucency, tricuspid regurgitation, ductus venosus with "a" wave reversal and abnormal facial angle. In the second trimester, sonographic markers of fetal aneuploidy (SMFA) are thickened nuchal fold, absent NB, ventriculomegaly, aberrant right subclavian artery, echogenic bowel, pyelectasia, echogenic intracardiac focus (EIF), short humerus, and short femur. Each of the above SMFA has a numerical likelihood ratio (LR).

Every fetus based on maternal age has a prior risk or background risk for aneuploidy. This risk gets altered into low risk or high risk based on the presence or absence of first and second trimester markers of aneuploidy.

Maternal biochemical analysis in first trimester—double marker test [β-human chorionic gonadotropin (β-hCG) and pregnancy-related plasma protein A] and in second trimester—triple marker test (α-fetoprotein, estradiol, and β-hCG) and quadruple marker test (β-hCG, estradiol, α-fetoprotein, and inhibin) will alter the above risk of aneuploidy and then arrive at a combined risk which is the best method of determining the risk for aneuploidy.

Cardiac anomaly detection qualifies as major abnormality and will increase the risk of aneuploidy irrespective of presence or absence of other abnormalities and hence will indicate requirement of karyotype analysis by invasive or noninvasive methods. Thus, detection of extracardiac anomalies and associated genetic/chromosomal abnormalities in a case of cardiac anomaly helps in counseling regarding severity of the situation and further course of action.

The following text will help the readers to understand the spectrum of disorders that may be associated with cardiac anomalies.

CARDIAC ANOMALIES AND GENETIC SYNDROMES

Cardiac anomalies can be isolated or can be associated with aneuploidy and when

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associated with aneuploidy they can be part of a genetic syndrome. If a cardiac anomaly is detected, then a detailed targeted imaging of the fetus is advised to look for any other anomalies. The list of genetic syndromes associated with cardiac anomalies is exhaustive and hence, it is not important to know about all the genetic and chromosomal syndromes, but it is important to understand certain principles to be followed when a cardiac anomaly is identified:

- History taking is very important.
  - Detailed medical history of both parents and family for any autosomal dominant or recessive conditions
  - History of consanguinity
  - Sibling history of cardiac anomaly or genetic disorder
  - History of teratogenic or anticonvulsant medications
- Targeted imaging for fetal anomalies scan increases the detection rate of anomalies
- Maternal biochemistry helps in risk analysis and increases detection rate of aneuploidy
- Chorionic villus sampling, amniocentesis, cordocentesis with fluorescence in situ hybridization (FISH) for 22q11 deletion, karyotype will be helpful in identifying aneuploidies.

The common genetic syndromes are:

- Holt–Oram syndrome
- Short rib polydactyly syndrome
- Ellis–van Creveld syndrome
- VACTERL syndrome
- Thrombocytopenia-absent radius (TAR) syndrome
- Cornelia de Lange syndrome
- Campomelic dysplasia
- Smith–Lemli–Opitz syndrome
- Noonan's syndrome
- Heterotaxy syndromes
- Anticonvulsant syndrome
- Goldenhar syndrome/hemifacial syndrome
- CHARGE syndrome
- Tuberous sclerosis
- Marfan's syndrome
- Alagille syndrome
- Williams syndrome

Holt–Oram Syndrome

This condition is inherited as autosomal dominant fashion with high penetrance and variable expression.

Some cases are caused by mutations on the *TBX5* gene on 12q2 chromosome. Atrial septal defect (ASD) occurs in 90% of cases. Hence, it is also called atriodigital dysplasia. Conduction disorders, ventricular septal defect (VSD), aortic coarctation, truncus arteriosus, patent ductus arteriosus (PDA), and mitral valve defects can occur.

Skeletal anomalies include thumb aplasia, hypoplasia, triphalangeal thumb, radial dysplasia, hypoplasia, radial fusion, phocomelia, clavicle, humerus, and ulnar hypoplasia.

Short Rib Polydactyly Syndrome

These are a group of autosomal recessive lethal skeletal dysplasia distinguished by the presence of thoracic hypoplasia, short ribs, short limbs, postaxial polydactyly **(Fig. 1)**, gastrointestinal anomalies, and other internal organ anomalies.

The potentially surviving conditions are:

- Thoracic asphyxiating syndrome (Jeune's syndrome)
- Ellis–van Creveld syndrome (chondroectodermal dysplasia)

A wide variety of cardiac malformations may occur including atrioventricular septal defect (AVSD), persistent left superior vena cava (LSVC), situs inversus, transposition of

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Fig. 1: Postaxial polydactyly.

the great arteries (TGA), aortic coarctation, hypoplastic ventricle, and VSD. Recurrence risk is about 25%.

Ellis–van Creveld Syndrome (Chondroectodermal Dysplasia)

This condition is inherited as an autosomal recessive fashion and is characterized by short limbs, short ribs, postaxial polydactyly, cardiac renal anomalies, dysplastic nails, and teeth. Characteristic tetrad includes chondrodysplasia, ectodermal dysplasia, postaxial polydactyly, and congenital cardiac anomalies. It is caused by mutation on 4p16. Cardiac anomalies include ASD, VSD, common atrium, AVSD, and situs inversus.

VACTERL Syndrome

These are a group of disorders arising due to defect in mesodermal development in the primitive streak. It is a mitochondrial disorder with a recurrence rate of 1–50%. It is inherited as an autosomal recessive and X-linked disorder. Conditions with a spectrum of disorders, such as vertebral anomalies **(Figs. 2A to H)**, anal atresia, cardiac anomalies, tracheoesophageal fistula, esophageal atresia, radial dysplasia, and renal and limb **(Figs. 3A to E)** anomalies have been grouped under the name VACTERL. Cardiac anomalies are a frequent occurrence.

Thrombocytopenia-absent Radius Syndrome

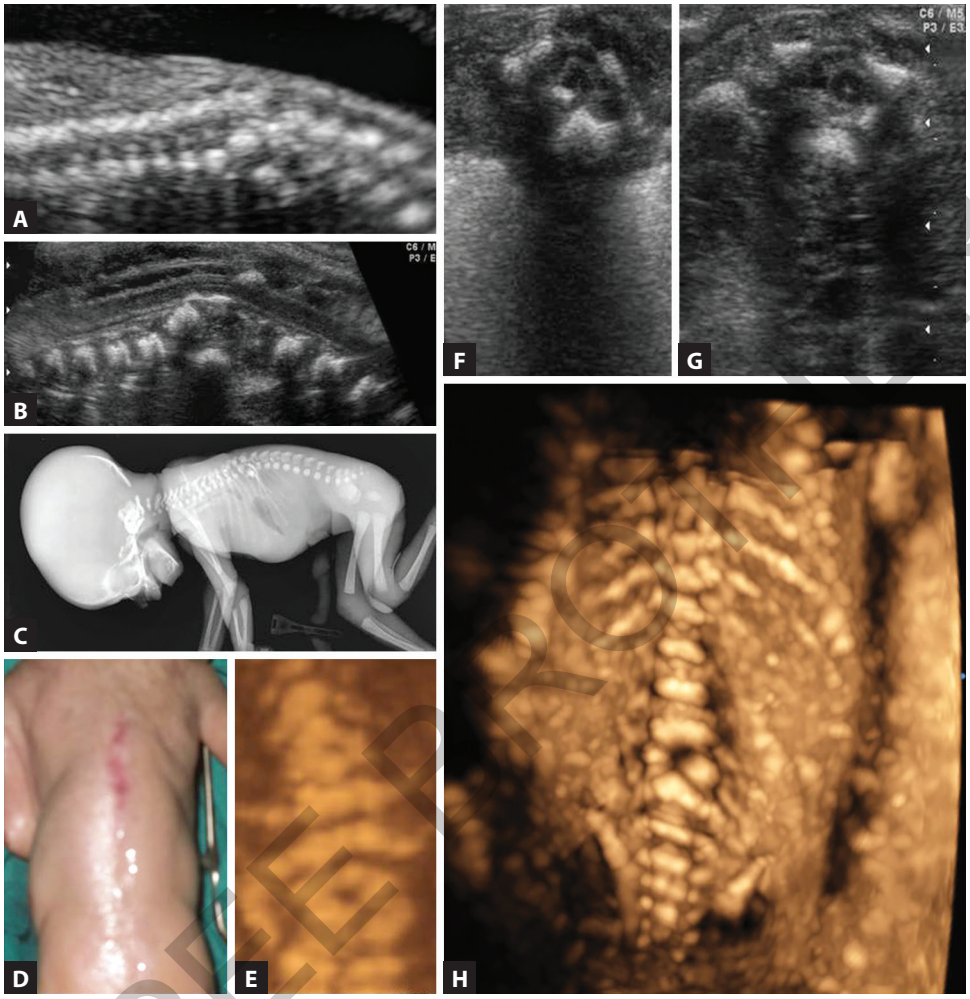
It is an autosomal dominant condition with variable penetration. This is characterized by the absence of both radius and preservation of both thumbs and thrombocytopenia. In 50% of case, there can be absent ulna or humerus. In 40% of cases, there may be lower limb anomalies. In 23% of cases, there may be renal anomalies. Cardiac anomalies are seen in 20–33% cases and can be tetralogy of Fallot (TOF), ASD (>50%), VSD, AVSD, and PDA in 15% cases.

Cordocentesis may detect thrombocytopenia. Intrauterine platelet transfer prior to delivery can be helpful. Chromosomal analysis may show 22q11 deletion.

Cornelia De Lange Syndrome

It is inherited as an autosomal dominant or X-linked disorder. There is a 50% chance of passing the disorder to the offspring. This condition is characterized by limb abnormalities, intrauterine growth restriction (IUGR), hearing loss, microcephaly, hirsutism, synophrys, thick eyelashes, low frontal hairline, anteverted nostrils, long philtrum, thin lips, retrognathia, and low ears. It is an autosomal dominant condition and 99% of cases are sporadic. Upper limb anomalies include oligodactyly, thumb and first metacarpal hypoplasia, clinodactyly of fifth finger, aplasia of ulna, and elbow fusion. Other features include diaphragmatic hernia, nuchal webbing, duodenal atresia, renal dysplasia, cleft palate, and genital anomalies. Cardiac anomalies include VSD, ASD, pulmonary stenosis, TOF, mitral

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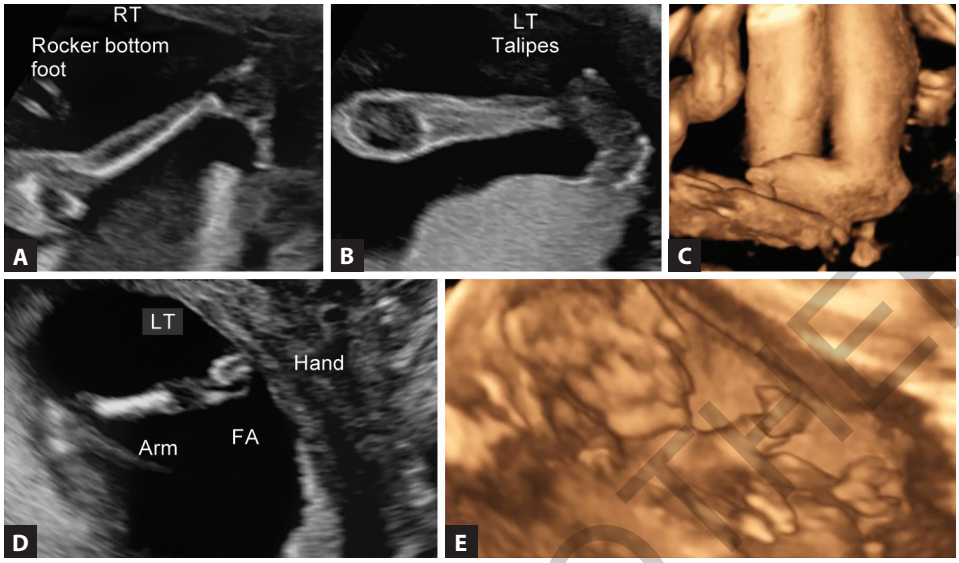
Figs. 2A to H: *Fetus with hemivertebrae:* (A) Fetal ultrasound showing spinal hemivertebrae; (B) Abortus ultrasound showing hemivertebrae; (C) Abortus X-ray showing hemivertebrae; (D) Abortus picture of the spine; (E) Three-dimensional (3D) ultrasound of spine; (F and G) Axial ultrasound of abortus showing the same; and (H) 3D of lumbar spine showing hemivertebrae of L4 vertebral body.

atresia, coarctation of aorta, AVSD, and single ventricle. Clues to the condition are oligodactyly, late-onset IUGR and microcephaly, and low pregnancy-associated plasma protein A (PAPP-A).

Campomelic Dysplasia

It is also called campomelic dwarfism. This condition is seen frequently and is caused by a severe defect in cartilage development causing bowing of femur and tibia, talipes, hypoplastic scapulae, 11 pairs of ribs, micrognathia, and a small chest. One-third has cardiac malformations including VSD, ASD, and TOF. Hypospadiasis and bifid scrotum can occur. Neonatal death occurs due to pulmonary hypoplasia. Heterozygous mutation of SOX9 located on 17q24 is the cause.

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Figs. 3A to E: *Limb anomalies:* (A) Rocker bottom foot with loss of plantar arch; (B) Talipes equinovarus; (C) 3D image showing talipes; (D) Radial ray anomaly—absent radius, short ulna, and ulna deviated in turned hand with hypoplastic wrist; and (E) 3D image showing same anomaly as D. (FA: forearm)

Cardiac Anomalies and Nuchal Edema/Hydrops

Accumulation of subcutaneous fluid may range from increase in nuchal translucency to nuchal fold to cystic hygroma to hydrops **(Figs. 4A to E)**. Hydrops is characterized by accumulation of fluid in causing ascites, pleural, or pericardial effusion. Cardiac conditions associated with nonimmune hydrops include tachyarrhythmias, congenital heart block, cardiomyopathy, myocarditis (coxsackievirus and cytomegalovirus). Associated genetic syndromes are:

Smith–Lemli–Opitz Syndrome

This condition is an autosomal recessive condition caused by a mutation in 7-dehydrocholesterol reduction gene on 11q13. This causes elevated levels of 7-dehydrocholesterol, which causes teratogenic effects. Polydactyly, toe syndactyly, thickened NT, cataract, renal anomalies, ambiguous male genitalia, and holoprosencephaly can occur. Cardiac anomalies include AVSD, ASD, TOF, PDA, aortic coarctation, hypoplastic left heart syndrome (HLHS), pulmonary stenosis, and tricuspid atresia. CVS can be used to detect 7-dehydrocholesterol reductase activity. Material urine testing can also detect the condition.

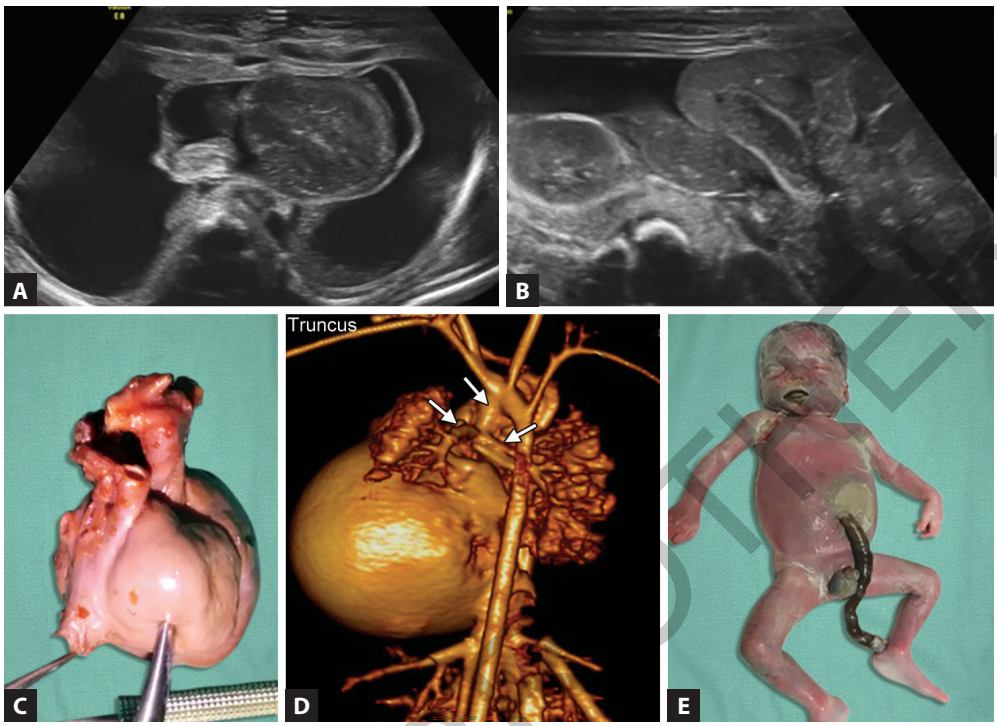
Noonan's Syndrome

This is an autosomal dominant condition caused by mutation in PTPN11. It is characterized by short stature, facial dysmorphism, webbed neck **(Fig. 5)**, and cardiac anomalies. Cardiac anomalies include left ventricular hypertrophy, dysplastic pulmonary valve, pulmonary stenosis, and ASD. Hydrops, short femur, and cardiac anomaly should alert the condition.

Heterotaxy and Isomerism

These are conditions associated with characteristic cardiac and abdominorespiratory anomalies and also referred to as isomerism

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Figs. 4A to E: *Hydrops:* (A and B) Abortus sonography showing bilateral pleural effusion, pericardial effusion, and ascites as signs of hydrops; (C) Cardiac autopsy showing cardiomegaly with truncus atreriosus; (D) Abortus CT ventriculography shows dilated heart with truncus arteriosus on CT VRT with single cardiac outlet giving rise to both aorta and pulmonary arteries; and (E) Abortus picture shows swollen fetus due to cardiac failure and hydrops. (CT: computed tomography; VRT: volume rendering technique)

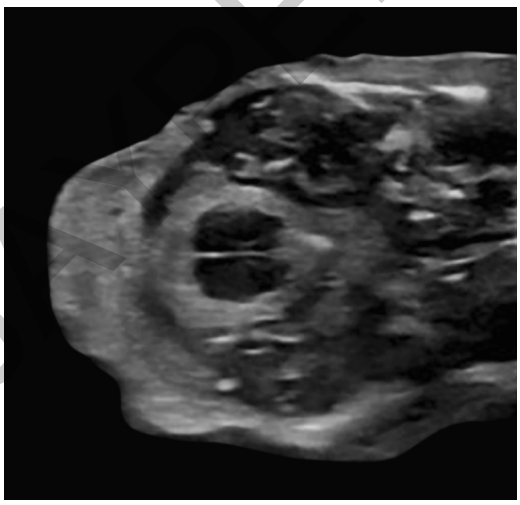


Fig. 5: Thickened nuchal fold/webbed neck.

of the atrial appendages. It is defined as an abnormal arrangement of thoracic and abdominal organs from the normal situs solitus **(Fig. 6)** arrangement caused by a disruption of the left right arrangement during embryonic development. Cardiac anomalies are a major component. There can be paired right atria resulting in right atrial isomerism or paired left atria resulting in left atrial isomerism.

Right Atrial Isomerism

This condition **(Fig. 7)** has paired right-sided viscera, bilateral trilobed lungs, tracheal atresia and enlarged obstructed echogenic lungs,

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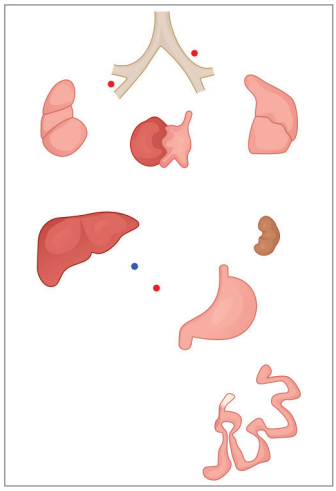


Fig. 6: Diagrammatic representation of situs solitus.

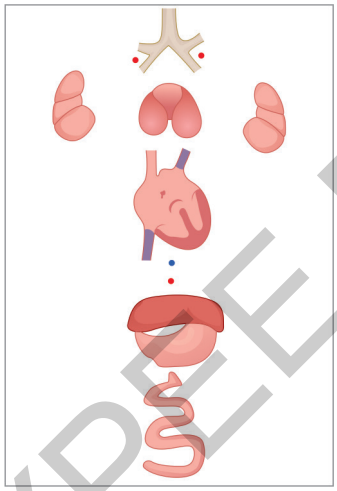


Fig. 7: Diagrammatic representation of right-sided isomerism.

esophageal atresia **(Figs. 8A to M)**, total anomalous pulmonary venous connection (TAPVC), TGA, pulmonary atresia, midline liver, central stomach, inferior vena cava (IVC) anterior to aorta in same sagittal plane, and absent spleen **(Figs. 8N to S)**. Asplenia is associated with serious cardiac anomalies (Ivemark syndrome). CNS and renal anomalies can occur.

Left Atrial Isomerism

This condition **(Fig. 9)** has bilateral bilobed lungs, malpositioned stomach, and polysplenia.

Atrioventricular septal defect, IVC interruption with azygos continuation **(Figs. 10A to J)**, and bilateral superior vena cava (SVC) are common. About 5–10% sibling risk is implied in nonconsanguinity.

Cardiac anomalies in heterotaxy syndromes include dextrocardia, single atrium/ASD, single ventricle/VSD, TGA, common AV valve, total or partial anomalous pulmonary return, endocardial cushion defect, aortic coarctation, pulmonary atresia, and double outlet right ventricle (DORV).

Teratogens

Women taking anticonvulsants have a 2–7-fold increase in risk for malformations.

Neural tube defects **(Figs. 11A and B)**, genital, cardiac, and musculoskeletal anomalies may occur. VSD, ASD, pulmonary stenosis, and aortic stenosis may occur.

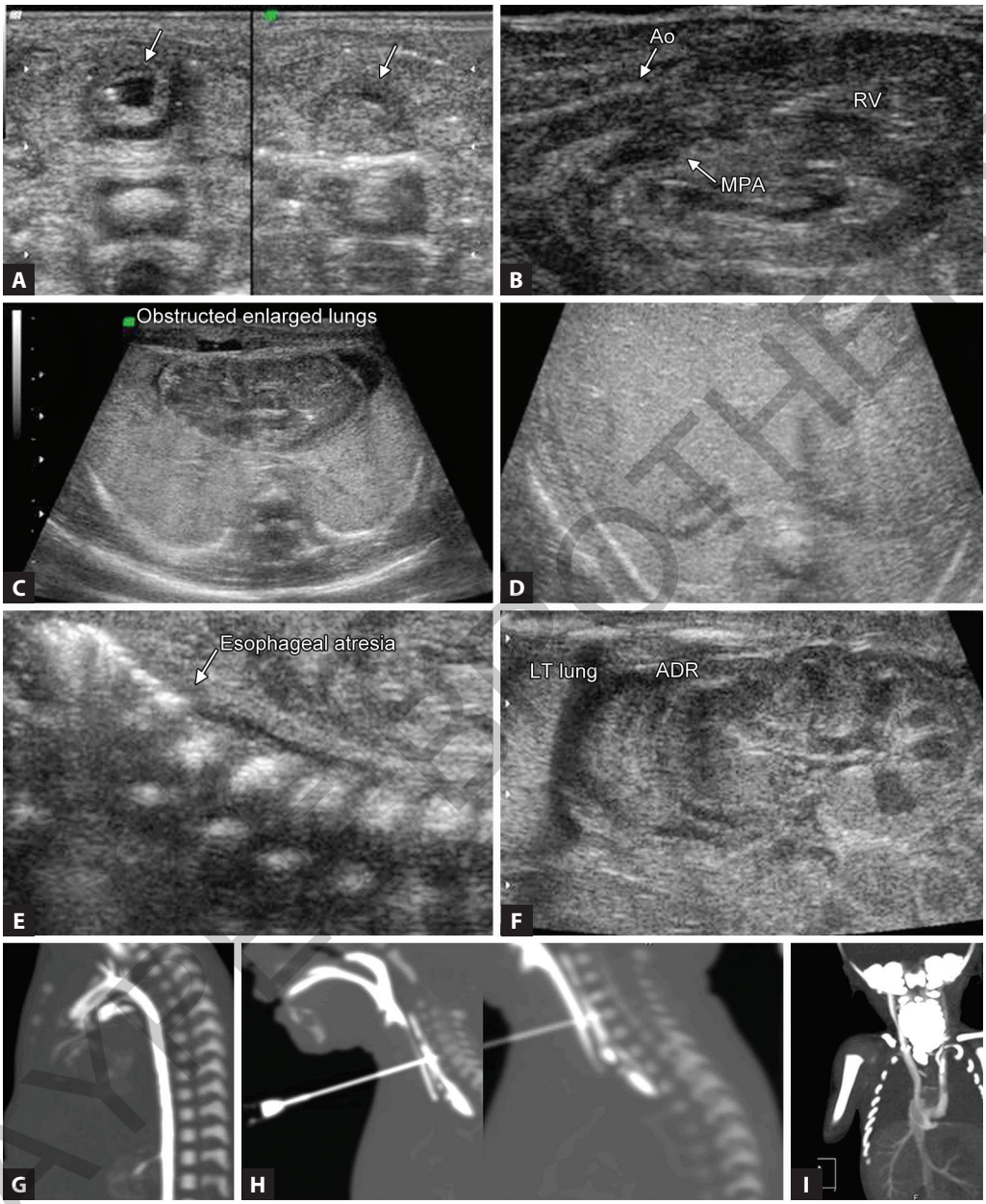
Goldenhar Syndrome/Hemifacial Microsomia

This condition is also called facio-auriculo-vertebral syndrome. It involves anomalies of the first and second branchial arches. Asymmetric facial deformities include ear deformities, preauricular tags **(Figs. 12A and B)**, ear dystopia, epibulbar dermoids, upper eyelid colobomas, facial clefting, and hemifacial microsomia. Vertebral, thoracic, and renal anomalies may occur. Cardiac anomalies include TOF and VSD. Unilateral microphthalmia **(Figs. 13A to D)**, orbital hypoplasia, and cleft lip **(Figs. 14A to D)** must alert the condition.

CHARGE Syndrome

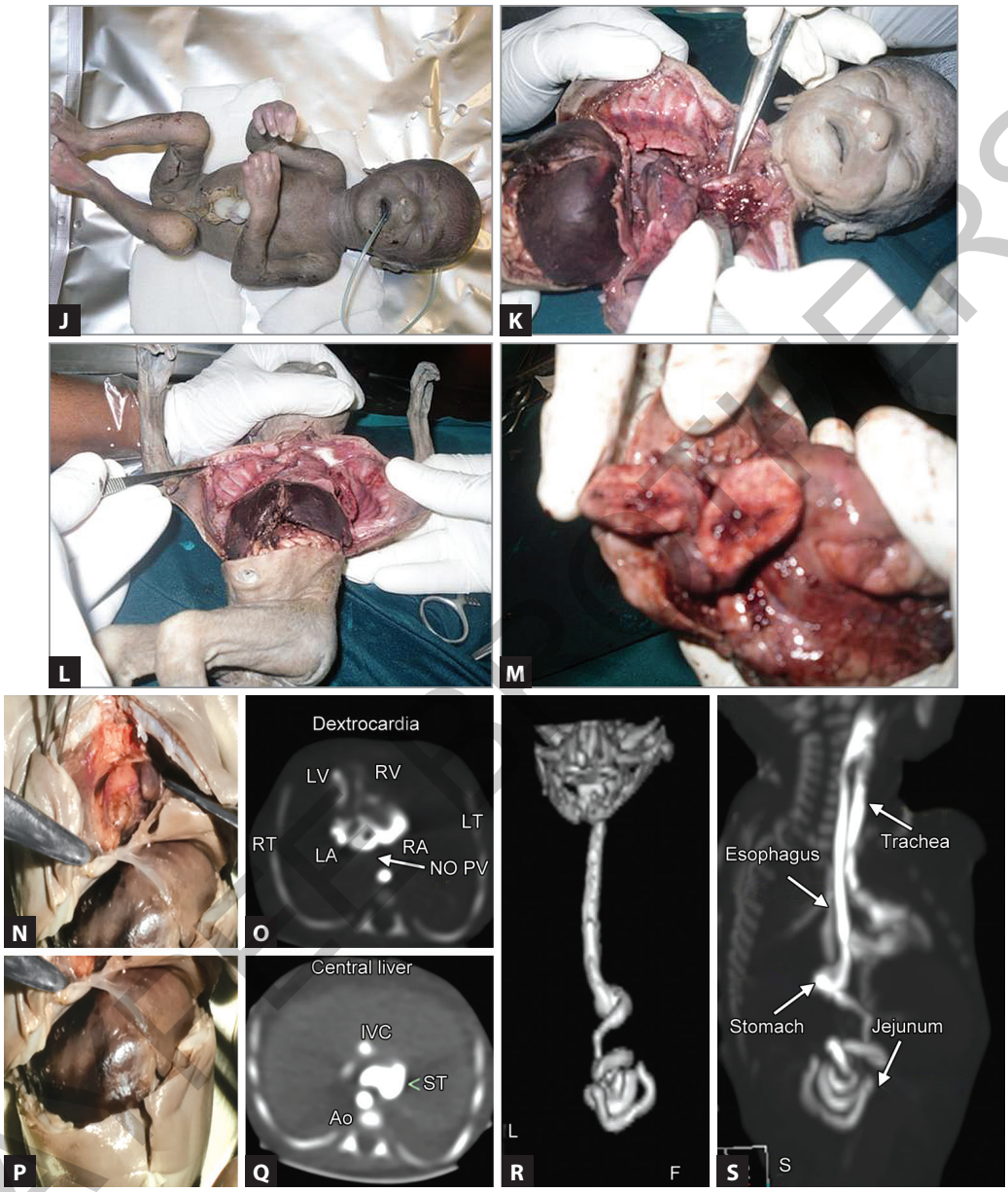
This condition encompasses coloboma of iris or retina, heart defects, atresia of choanae,

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Figs. 8A to I: Right-sided isomerism. (A to F) Ultrasound of abortus with right atrial isomerism. (A) Tracheal stenosis; (B) DORV showing anterior aorta and posterior pulmonary artery arising from RV; (C) Echogenic enlarged lungs; (D) Central liver and absent spleen; (E) Esophageal atresia with blind ending air column; (F) Absent spleen in left subdiaphragmatic area; (G to I) CT pictures of the abortus. (G) Contrast injected into right ventricle of abortus shows both aorta and pulmonary artery filling from RV. Note: Transposed anterior aorta and posterior pulmonary artery; (H) CT tracheogram and esophagogram of abortus—contrast injected into trachea and esophagus show blind ends—atresia; and (I) Contrast injected into right atrium shows two SVC and one IVC. (ADR: adrenal gland)

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Figs. 8J to S: (J to M) Dissection of abortus with right atrial isomerism. (K) Esophageal atresia; (L) Central liver and absent spleen; and (M) DORV with transposed great vessels. (N to S): Right-sided isomerism. (N) Autopsy showing dextrocardia; (O) CT ventriculogram of the abortus showing cardiac apex pointing to the right, absent pulmonary veins with smooth posterior wall of left atrium suggesting total anomalous pulmonary venous connection; (P) Autopsy showing central liver; (Q) Axial CT showing central liver, central stomach, IVC anterior to aorta (Ao); and (R and S) CT gastrogram showing normal esophagus, midline gastroesophageal junction, small midline stomach and duodenum, abnormal duodenojejunal junction, and malrotated jejunum. (CT: computed tomography; DORV: double outlet right ventricle; IVC: inferior vena cava; MPA: main pulmonary artery; RV: right ventricle; SVC: superior vena cava)

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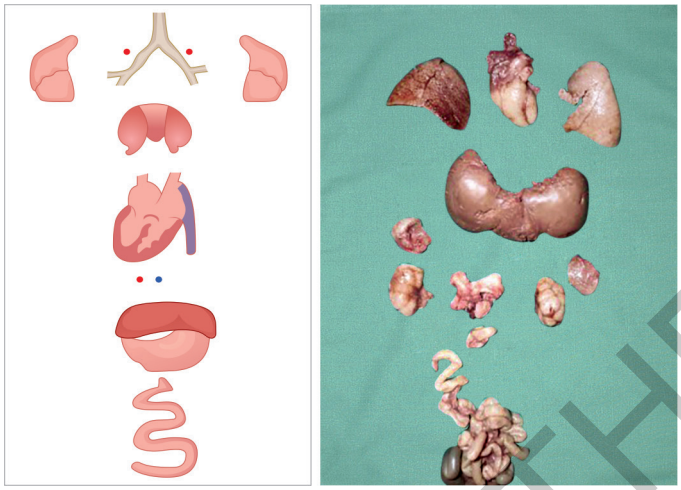


Fig. 9: Left-sided isomerism, diagrammatic representation with autopsy specimen.

retardation of mental and somatic development, genital anomalies, ear abnormalities, and deafness. Cardiac anomalies include conotruncal malformation, TOF, AVSD, PDA, outflow anomaly, arch anomalies, and VSD.

Tuberous Sclerosis

This condition has a prevalence of 1:6,000. It is inherited as autosomal dominant fashion. Many cases are sporadic. Changes occur in *TSC1* and *TSC2* gene of 9q34 and 16p. Features include depigmented macules, facial angiofibromas, ungual fibromas, cranial subependymal nodules, renal angiomyolipomas, renal cysts, and cardiac rhabdomyomas. Cardiac rhabdomyomas grow till 32 weeks and stabilize with excellent resolution postnatally. Fetal magnetic resonance imaging (MRI) may be helpful to detect subependymal nodules **(Figs. 15A to F)**.

Marfan's Syndrome

This condition comprises connective tissue disorders and cardiac malformations.

It is an autosomal dominant multisystem disorder. Abnormalities are expressed in musculoskeletal system, ocular system, and cardiovascular system predominantly.

The gene for fibrillin undergoes mutation. Cardiovascular abnormalities include aortic root dilatation and mitral valve prolapse. This condition is seldom diagnosed antenatally.

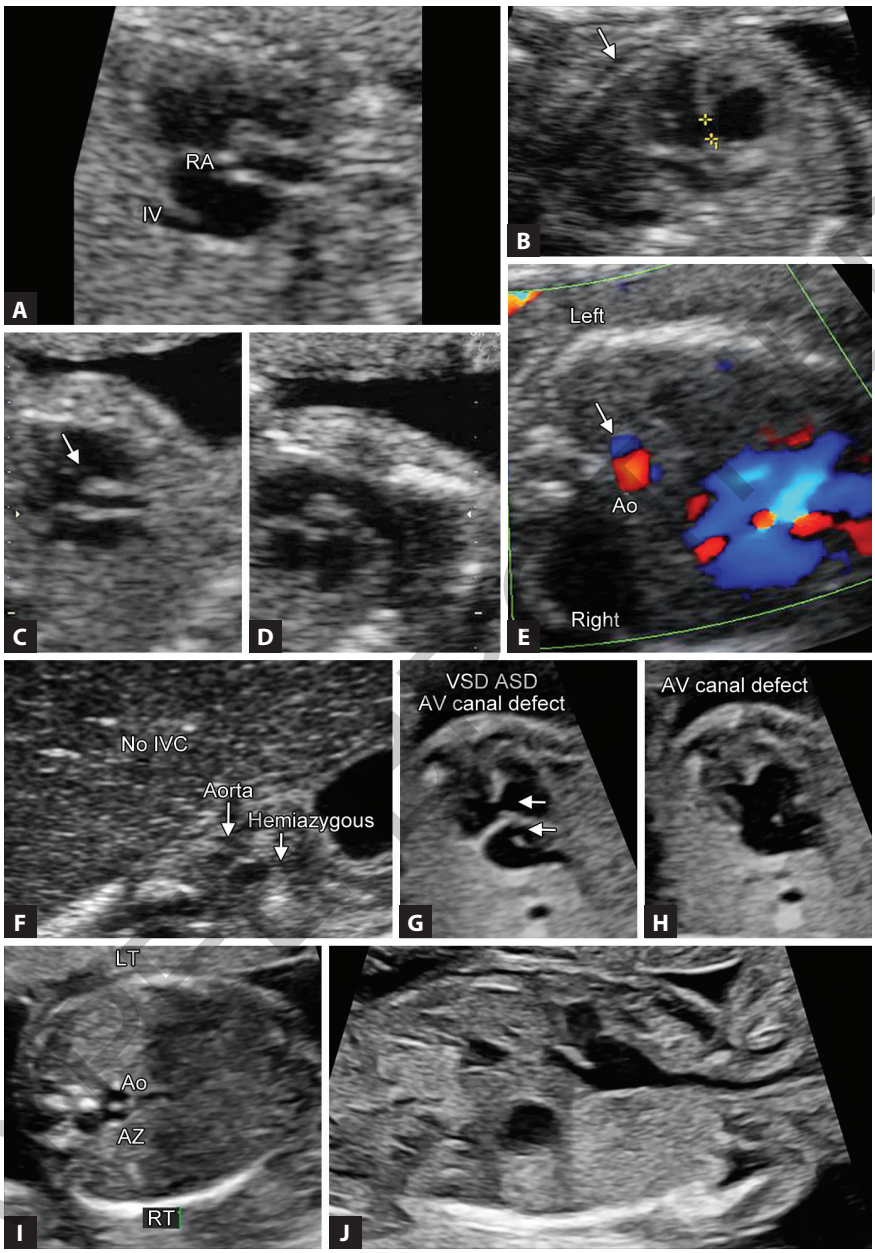
Alagille Syndrome

This condition is an autosomal dominant disorder with intrahepatic cholestasis, facial abnormalities (prominent forehead, deep set eyes, and long nose), ocular posterior embryotoxon, and hemivertebrae. Cardiac anomalies include TOF and peripheral pulmonary artery stenosis. Mutation or deletion of *Jagged 1* gene is observed.

Williams Syndrome

This condition is a contiguous gene deletion syndrome with 1–2 megabase deletion on long arm of chromosome 7. This results in defective elastin. Cardiac anomaly is characteristically supravalvular aortic stenosis. Other abnormalities include periorbital fullness, short anteverted nose **(Fig. 16)**, and long philtrum.

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Figs. 10A to J: Left atrial isomerism: (A to F) Fetal echo of left-sided isomerism. (A) IVC interruption; (B) VSD with mesocardia; (C) VSD in long-axis view with small size aorta; (D) RVOT view showing normal-sized pulmonary artery; (E) Hemiazygos vein to the left of aorta and absent IVC; (F) Absent IVC and hemiazygos vein to the left of aorta; (G and H) Atrioventricular septal defect (AVSD) in left-sided isomerism. (G) Single atrioventricular (AV) valve, ASD, and VSD seen in ventricular systole; (H) Boomerang-shaped Crux and AV valve in systole, in ventricular diastole there is complete absence of crux with empty heart sign; and (I and J) Interrupted IVC and azygous at the same level as aorta to its right. (IVC: inferior vena cava; RVOT: right ventricular outflow tract; VSD: ventricular septal defect)

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