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FETAL ECHOCARDIOGRAPHY: INDICATIONS

Indications for Fetal Echocardiography

1. Familial risk factors

- *History of congenital heart disease:*

  Previous sibling

  Paternal

- *Mendelian syndromes that include congenital heart disease:*

  Noonan

Indications for Fetal Echocardiography

2. Maternal risk factors

A. In vitro fertilization
B. Congenital heart disease
C. Cardiac teratogen
D. Maternal metabolic disorders
   - Diabetes mellitus
   - Phenylketonuria

Indications for Fetal Echocardiography

3. Fetal risk factors

- Extracardiac anomalies
  - Chromosomal
  - Anatomic
  - Increased nuchal fold thickness
- Fetal cardiac arrhythmia
  - Irregular rhythm
  - Tachycardia (> 180 bpm) in absence of amnionitis
  - Fixed bradycardia

Indications for Fetal Echocardiography

- Nonimmune hydrops fetalis
- Abnormal fetal situs
- Suspected fetal heart malformation on screening ultrasound
- Lack of reassuring four-chamber view during basic obstetric scan

- *The most common indication for performing a fetal echocardiogram:*
- is a *family history of congenital heart disease*

- *The risk of occurrence* for a fetus depends on :

1. the type of lesion
2. the relationship of the fetus to the affected relative

- *The risk of CHD in a fetus with an affected sibling*:
- is approximately 2% to 4%

- *If two or more siblings are affected:*

- risk increases to about 10%

- *When the mother of the fetus is the affected relative:*
- the risk of a heart defect is also approximately 10% to 12%

- *If the affected relative is the father:*
- The risk is lower

NOTE

- If congenital heart disease does recur in families:
- it is not limited to the same type of defect

Risk of Occurrence for Any Congenital Heart Defect in Siblings

Defect	Suggested Risk	
	If One Sibling Affected	If Two Siblings Affected
Aortic stenosis	2	6
Atrial septal defect	2.5	8
Atrioventricular septal defect	3	10
Coarctation of the aorta	2	6
Ebstein anomaly	1	3
Endocardial fibroelastosis	4	12
Hypoplastic left heart syndrome	2	6
Pulmonary atresia	1	3
Pulmonary stenosis	2	6
Tetralogy of Fallot	2.5	8
Transposition of the great arteries	1.5	5
Tricuspid atresia	1	3
Truncus arteriosus	1	3
Ventricular septal defect	3	10

Offspring Occurrence Risk for Congenital Heart Defects Given One Affected Parent (%)

Defect	Suggested Risk (%)	
	Father Affected	Mother Affected
Aortic stenosis	3	13–18
Atrial septal defect	1.5	4–4.5
Atrioventricular septal defect	1	14
Coarctation of the aorta	2	4
Pulmonary stenosis	2	4–6.5
Tetralogy of Fallot	1.5	2.5
Ventricular septal defect	2	6–10

Cardiac teratogens

- *Exposure to known cardiac teratogens:*

- *also increases* fetal risk for a heart defect

Cardiac teratogens

- *The specific risk of occurrence varies with:*
- The length and types of exposure
- with the specific substance involved

Substances Associated with Congenital Heart Disease

Substance	Associated Congenital Heart Disease
Alcohol	Atrial septal defect, ventricular septal defect, interrupted aortic arch, coarctation, tetralogy of Fallot, pulmonary stenosis, double-outlet right ventricle, dextrocardia
Amantadine	Single ventricle, pulmonary atresia
Amphetamine	Ventricular septal defect, atrial septal defect, transposition of the great arteries
Azathioprine	Pulmonary stenosis
Barbiturates	Interrupted aortic arch, coarctation
Cannabis	Ventricular septal defect
Carbamazepine	Atrial septal defect
Chlordiazepoxide	Congenital heart disease (unspecified)
Codeine	Congenital heart disease (unspecified)
Cortisone	Ventricular septal defect, coarctation
Cyclophosphamide	Tetralogy of Fallot
Cytarabine	Tetralogy of Fallot
Daunorubicin	Tetralogy of Fallot
Dextroamphetamine	Atrial septal defect
Diazepam	Congenital heart disease (unspecified)
Dilantin (hydantoin)	Atrial septal defect, ventricular septal defect, interrupted aortic arch, coarctation, pulmonary stenosis, aortic stenosis
Indomethacin	Ductal constriction
Lithium	Ebstein anomaly, tricuspid atresia, atrial septal defect, mitral atresia, dextrocardia
Methotrexate	Dextrocardia

Indomethacin	Ductal constriction
Lithium	Ebstein anomaly, tricuspid atresia, atrial septal defect, mitral atresia, dextrocardia
Methotrexate	Dextrocardia
Oral contraceptives	Congenital heart disease (unspecified)
Paramethadione	Tetralogy of Fallot
Penicillamine	Ventricular septal defect
Primidone	Ventricular septal defect, interrupted aortic arch, coarctation
Progesterone	Ventricular septal defect, tetralogy of Fallot, truncus arteriosus
Quinine	Congenital heart disease (unspecified)
Retinoic acid (Accutane)	Ventricular septal defect, interrupted aortic arch, coarctation, tetralogy of Fallot, truncus arteriosus, double-outlet right ventricle, pulmonary atresia
Thalidomide	Ventricular septal defect, transposition of the great arteries, truncus arteriosus, tetralogy of Fallot, double-outlet right ventricle, pulmonary atresia, atrial septal defect
Trifluoperazine	Transposition of the great arteries
Trimethadione	Ventricular septal defect, transposition of the great arteries, tetralogy of Fallot, hypoplastic left heart syndrome, double-outlet right ventricle, pulmonary atresia, truncus arteriosus, atrial septal defect, aortic stenosis, pulmonary stenosis
Valproic acid	Ventricular septal defect, coarctation, interrupted aortic arch, tetralogy of Fallot, hypoplastic left heart syndrome, aortic stenosis, atrial septal defect, pulmonary stenosis
Warfarin (Coumadin)	Congenital heart disease (unspecified)

Chromosome Abnormalities and Syndromes Associated with CHD

- *Achondroplasia:*
- Interrupted aortic arch, coarctation

- *Asymmetric crying face:*
- Tetralogy of Fallot, ventricular septal defect

- *CHARGE:*
- Including:
- coloboma of the eye, heart anomaly, choanal atresia, retardation, and genital and ear anomalies syndrome

- AVSD, COA, VSD, ASD, truncus arteriosus, DORV, TOF, right aortic arch

- *Factor V deficiency:*

- Atrial septal defect, ventricular septal defect

- *VACTERL:*

- vertebral abnormalities, anal atresia, cardiac abnormalities, tracheoesophageal fistula or esophageal atresia, renal agenesis and dysplasia, and limb defects syndrome

- HLHS, ventricular septal defect

- *Velocardiofacial syndrome:*

1. Ventricular septal defect
2. tetralogy of Fallot
3. right aortic arch

- *William syndrome:*

1. Aortic stenosis
2. pulmonary stenosis
3. ventricular septal defect
4. atrial septal defect
5. interrupted aortic arch
6. mitral regurgitation
7. tetralogy of Fallot
8. coarctation

Incidence (%) of Associated Congenital Heart Defects Occurring with Extracardiac Malformations in Infants

System or Lesion	Frequency of Congenital Heart Disease (%)
Central Nervous System	
Hydrocephalus	4.5–14.8
Dandy-Walker malformation	2.5–4.3
Agenesis of the corpus callosum	14.9
Meckel-Gruber syndrome	13.8
Gastrointestinal	
Tracheoesophageal fistula	14.7–39.2
Duodenal atresia	17.1
Jejunal atresia	5.2
Anorectal anomalies	22
Imperforate anus	11.7
Ventral Wall	
Omphalocele	19.5–32
Gastroschisis	0–7.7
Diaphragmatic Hernia	9.6–22.9
Genitourinary	
Renal agenesis (bilateral)	42.8
Renal agenesis (unilateral)	16.9
Horseshoe kidney	38.8
Renal dysplasia	5.4
Ureteral obstruction	2.1

Chromosomal abnormalities

- *occur in:*
- 13% of live-born infants with CHDs
- 35% in the fetus with a congenital heart abnormality

- the specific type and occurrence risk of a CHD varies depending on:

- The chromosomal abnormality

- *Some abnormal karyotypes:*
- have a relatively low association with heart defects

- *whereas others:*
- such as trisomy 21
- associated with a 40% to 50% occurrence

- *The most striking relationship:*
- trisomy 13 and trisomy 18
- in which the association with congenital heart abnormalities is almost 100%

- the list of abnormal karyotypes and syndromes associated with heart defects is extensive

maternal conditions

- *also carry an inherent* risk to the fetus

In vitro fertilization

- *Infants born of assisted reprodactive technology:*
- Are more likely to be born preterm, LBW and SGA
- With IVF:
- There is 4 fold increase in CHD

Maternal obesity

- Defined as BMI greater or equal to 30 kg/m2
- Increased risk for CHDs although reletively small of about 1.18 fold
- Majority of cases :

1. VSD
2. ASD

diabetic mothers

- fivefold to eighteen fold increased risk of CHD compared with control subjects
- Diabetes, as an indication for fetal echocardiography

- *an overall increase in type 2 diabetes:*
- secondary to an increase in obesity and other medical problems
- ***Congenital cardiac defects most often associated with diabetes:***

1. transposition of the great arteries
2. truncus arteriosus
3. tetralogy of Fallot

Maternal phenylketonuria

- has a reported risk of 12% to 16% to the fetus of having a congenital heart defect

maternal collagen vascular disease

- Complete heart block in the fetus is associated with maternal SLE
- *Circulating antinuclear antibodies of the SSA or SSB type:*
- damage the developing conduction tissue

- *These fetuses may also be at risk for:*

- Premature closure of the ductus arteriosus

- as a result of maternal steroid treatment.

Maternal infection

- associated with heart defects in the fetus

- Commonly:

1. dilated cardiomyopathy
2. hypertrophic cardiomyopathy

Fetal risk factors

Monochromic placentation

- Associated with increased risk for CHDs
- Even after excluding TTTS
- Overral risk is about 9.1%
- *If one twin is affected:*
  The risk for the other one is 26.7%

Monochorionic placentation

- *The most common CHD:*

- Without TTTS:
- Is VSD

- With TTTS:
- Is PS and ASDs

extracardiac anomalies

- Found during an obstetrical sonographic examination
- warrants a fetal echocardiogram

- *Some extracardiac malformations:*
  - carry a low risk of associated CHD
  - whereas for others the risk is high

The overall incidence of extracardiac malformations in children identified as having CHD ranges from 25% to 45%

- atrioventricular (AV) septal defects:
- associated with extracardiac defects in more than 50% of cases

- atrial septal defects, ventricular septal defects, tetralogy of Fallot, and cardiac malposition

- are associated with extracardiac malformations in about 30% of cases

A suspicion of a *structural or rhythm bnormality*

- seen in the fetal heart on a routine obstetrical examination
- is another indication for a formal fetal echocardiogram

Important note

Up to 90% of congenital heart disease occurs in unselected "normal" obstetric patients

note

- *routine obstetrical scanning:*

- should identify the majority of fetuses with heart lesions needing a formal fetal echocardiogram

nonimmune hydrops fetalis

- an indication for fetal echocardiography
- *In some cases:*
- structural heart disease
- the result of an arrhythmia

- *Hydrops fetalis:*

- associated with:

- structural heart disease in *13.7%*
- cardiac rhythm abnormalities in *10.4%*

Polyhydramnios

- *indication for* fetal echocardiography
- may not be caused directly by the heart defect
- *but is more likely related to associated defects in the fetus*
- *such as those that cause difficulty in swallowing or compression of the esophagus*

Polyhydramnios

- *Fetal echocardiogram referrals for fetuses with polyhydramnios:*
- decreased over years past
- usually only cases of significant polyhydramnios

increased nuchal translucency

- identified in the first trimester
- The frequency of CHD is increased

- *In a fetus with an increased nuchal translucency and a confirmed chromosome anomaly:*

- the percent risk is that associated with the specific anomaly

Increased nuchal translucency

- As an isolated finding:
- carries a risk of an associated heart defect of approximately 2% to 5%
- *This risk increases exponentially with an increase in nuchal translucency thickness.*

Increased nuchal translucency

- CHDs found in chromosomally normal fetuses with an increased nuchal translucency:
- *left-sided defects such as:*

1. HLHS
2. Coarctation of the aorta
3. aortic stenosis
4. aortic atresia

Increased nuchal translucency

- Theory:

- pathological examination of the heart and great vessels in these fetuses after termination:

- *show a greater degree of narrowing of the aortic isthmus than seen in normal fetuses*

Increased nuchal translucency

- narrowing could result in greater perfusion of the head and neck
- which in turn results in a transient increase in subcutaneous neck edema
- thus the increased nuchal translucency

note

- However:
- it should be kept in mind that a variety of heart defects are seen in the setting of increased nuchal translucency with normal chromosomes
- so this does not explain this finding in all affected fetuses

Other indicatins

- 1. cardiac echogenic focus in routine sonographic examination
- 2. pylectasis
- *3. echogenic fetal* bowel

note

- ***Congenital heart disease:***

- is about 8 times more common than trisomy 21 and 4 times more common than neural tube defects

- 2 conditions for which universal screening programs already exist!!!!

- *Congenital heart disease:*

- has serious implications for affected infants and their families

- ***abnormal cardiac results on prenatal sonography:***
- are the most frequent predictor of congenital heart disease
- routine screening by adequately trained
- sonographers should be available to all pregnant women

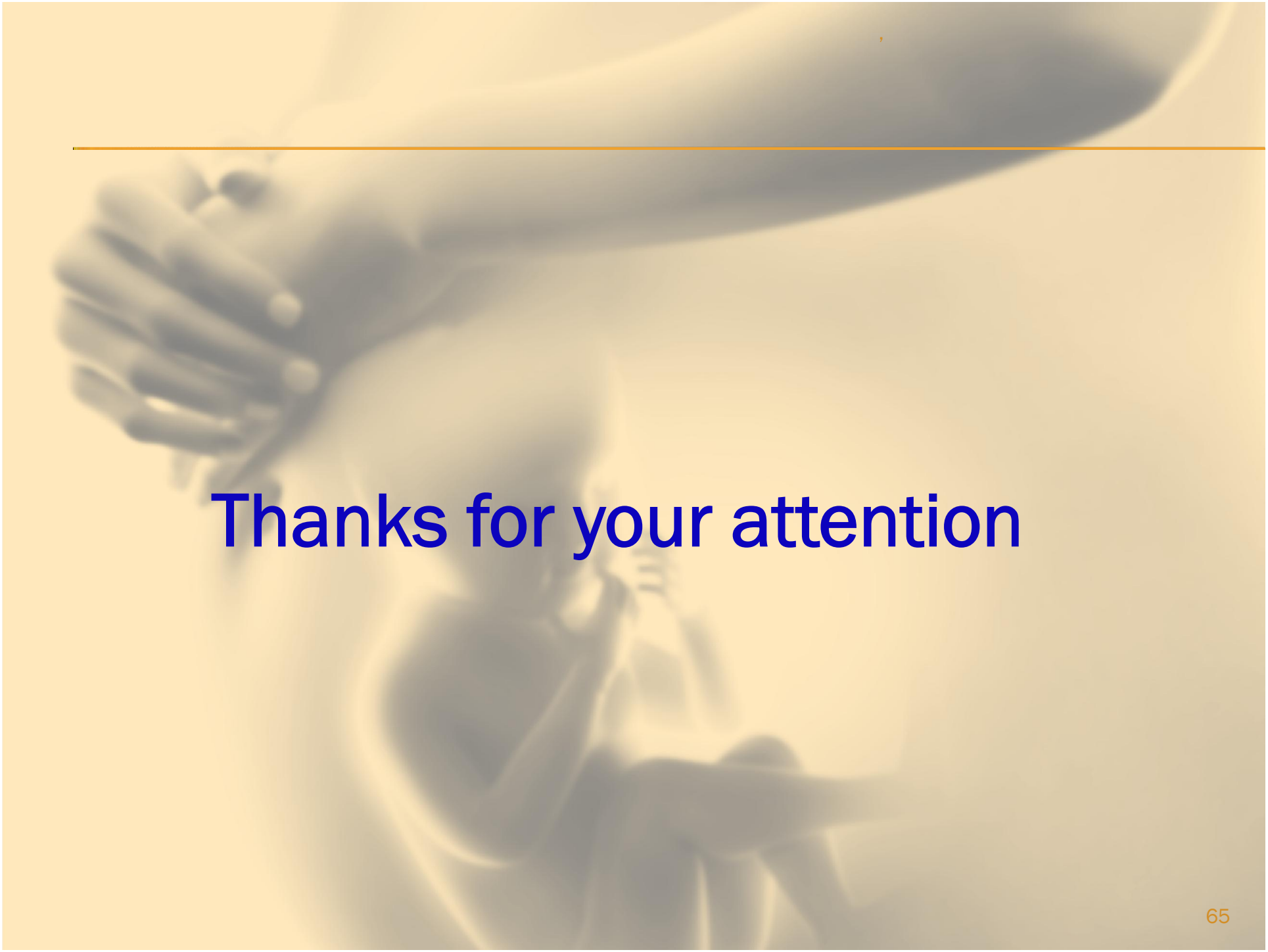
- Traditional risk factors:

- prompt discovery of relatively few cases of congenital heart disease on fetal echocardiography

- *careful assessment of the fetal heart during routine prenatal sonographic examination:*

- Has the greatest potential to improve the rate of prenatal detection of major cardiac defects

- Universal routine prenatal sonography should therefore be adopted as standard practice



Thanks for your attention