**Fetal Cardiology**

Fetuses have become our youngest cardiology patients

- Timeline for diagnosis and management of heart disorders has shifted to early in gestation
- As early as 12-14 weeks gestation heart defects can be detected
- Improved prenatal diagnostic screening and imaging
- Improved management of fetal heart and other disorders during pregnancy and at birth
- Fetal cardiac function and cardiovascular stability play an important role in fetal wellness
- Fetal circulation different from post-natal circulation
- Structural defects may progress in utero

**Prenatal Screening**

**NIPT (Noninvasive Prenatal Testing)**

- Screens for genetic conditions
  - 80% of rare diseases are genetic
- Maternal blood sample
  - No risk of miscarriage
  - Fetal genetic material – cfDNA
- Current NIPT screening includes:
  - Trisomy 21, Trisomy 18, Trisomy 13
  - Turner’s syndrome
  - 22q11.2 deletion, 1p36 deletion

**Invasive Prenatal Testing**

- Indicated when high suspicion
- Chorionic Villus Sampling (CVS)
- Amniocentesis
- Percutaneous Umbilical Blood Sampling (PUBS)
- Both with complete chromosome analysis and FISH analysis
- Risk of miscarriage or preterm labor and delivery
**Rationale for Prenatal Cardiac Screening**

### The Numbers
- Incidence of all birth defects is 3-5% of all live births. In addition, birth defects and chromosomal disorders result in a high percent of early miscarriages.
- Up to 50% of all conceptions, including 25% that are known pregnancies, end in a miscarriage.
- Congenital heart defects (CHD) are the most prevalent birth defect in newborns.
- CHD causes 20-30% of neonatal deaths.
- Up to 40% of all CHD are major heart defects.
- Up to 20-30% may have other anomalies.

### Critical Congenital Heart Disease

**Critical congenital heart disease (CCHD)** is defined as potentially life-threatening heart defects that require either surgical or catheter intervention in the first year of life.
- Includes heart defects that cause hypoxemia (ie, HLHS, TOF, TAPVR, d-TGA, truncus arteriosus, pulmonary or tricuspid atresia).
- Also includes other significant heart defects that sometimes but less consistently cause hypoxemia (ie, coarctation, IAA, single ventricle, Ebstein anomaly and DORV).

### Diagnosis of CCHD

Newborn physical exams in the nursery fail to detect more than 40% of CCHD.
- Newborns appear healthy while the ductus arteriosus remains open.
- No murmur or visible cyanosis is present.
- Need 3-5 grams of desaturated hemoglobin.
- Dark pigmented skin makes cyanosis detection difficult.
- Many newborns are roaming in with mothers so less observation by health care providers.
- Early hospital discharges.

**Rationale for Prenatal Cardiac Screening**

### More Numbers
- Incidence of a congenital heart defect (CHD) is about 1 in 120 live births (0.8%).
- 4.25 million live births in the US each year.
- 0.8% or 34,000 newborns will have CHD.
  - 40% or 13,600 will have at least significant CHD.
  - 25% or 8,500 will have critical CHD (CCHD).
  - This is the subset of newborns where early diagnosis is crucial in reducing morbidity and mortality. Each year a substantial number of babies are discharged home from the nursery with undiagnosed CCHD.

### Why screen for CCHD?
- Prior to the initiation of Pulse Oximetry Screening (POxS), 30% or about 2,500 of the 8,500 newborns with CCHD each year were missed at the time of discharge from the nursery. Even today, a significant number go undiagnosed.
- Thus the need for improved prenatal detection, improved newborn physical exam detection and the continuation of Pulse Oximetry Screening (POxS) of all newborns.
- For management of the newborn, prenatal diagnosis has the best chance of improving morbidity and mortality from CCHD.

### Critical Congenital Heart Defects (CCHD) can be diagnosed in one of the following ways:

- Prenatal imaging
- Newborn physical examination
**Diagnosis of CCHD**

- Pulse Oximetry Screening of the newborn
- At time of presentation in cardiovascular collapse
- Post mortem

**Best to worse times to diagnose CCHD**

- Prenatal Imaging
- Newborn Physical Exam
- Newborn Pulse Oximetry Screening
- At time of Presentation in Shock
- Post Mortem

**Prenatal Screening for CCHD**

**Fetal heart imaging**

- Introduced > 30 years ago but still challenging for most OB sonographers
- Overall detection rate of CCHD remains low
  - Across the USA the mean detection rate is < 50%
  - Major tertiary center have better detections rates
    - 60 – 80%
  - Fetal echocardiography has highest detection rate but still significant CCHD is being missed
    - > 90% when performed by a Fetal / Pediatric Cardiologist

**Why aren’t we better at detecting CCHD prenatally?**

- Pregnancies at high-risk for CHD usually get a fetal echocardiogram which has a high sensitivity
- However, > 90% of CHD occurs in low-risk pregnancies.
- Therefore, the burden to detect CHD falls on the obstetrical ultrasound which usually has low sensitivity to detect CHD.

**Prenatal detection rate of CCHD in the United States**

- No uniform standards for fetal heart imaging
- In some regions of the US, obstetricians do not even do fetal anatomy scans
- And, if an anatomy scan is done, imaging of the fetal heart is usually limited and may not include the minimal 4-chamber and outflow tract views
- Adequate imaging of the fetal heart can be time consuming

**Factors that influence the diagnostic capability of fetal heart imaging**

- Patient’s body habitus / obesity
  - Diabetes has an increased risk for CHD
- Ultrasound attenuation
  - Tissue penetration maybe limited in some women
  - Previous C-section
- Outdated equipment
- Imaging views obtained
  - 4-chamber, Outflow tracts, 3 Vessel, Situs/IVC
  - Know the normal; abnormalities get referred for a fetal echo
Prenatal Screening for CCHD

Factors that influence the diagnostic capability of fetal heart imaging (continued)

- Time of gestation
  - Ideal time is between 18 – 22 weeks gestation
  - Prior to this time, fetal heart is small and there is increased fetal motion
  - After this time, bones absorb ultrasound and shadowing effects imaging
- Experience of the OB ultrasonographer
  - Insufficient OB training in congenital heart disease
  - Do not need to know all heart defects but must recognize an abnormal
  - The more you do, the better you are

Prenatal Screening for CCHD

Obstetrical fetal anatomy ultrasound scans at 18-22 weeks gestation should include the following:

- Four chamber view of the fetal heart
- Outflow tracts of right and left ventricles
- Three vessel tracts (pulmonary artery, aorta and superior vena cava)
- Situs and inferior vena cava

Prenatal Diagnosis of CCHD

The Fetal Echocardiogram

- Directed, comprehensive anatomic and hemodynamic assessment of the fetal heart and cardiovascular system usually performed by a Pediatric / Fetal Cardiologist or MFM Specialist with expertise in congenital heart disease.
- Sensitivity for detecting all CHD including small septal defects and minor valve abnormalities is about 43%
- For major CHD the sensitivity is 90% with a specificity of 99.9%
- Fetal echocardiography is presently indicated in high or at risk pregnancies but only about 10% of CHD occurs in this subset of pregnancies

Prenatal Screening for CCHD

Factors that influence the diagnostic capability of fetal heart imaging (continued)

- Type of heart defect
  - 50% detection rate
  - Hypoplastic left heart syndrome (HLHS)
  - Ebstein’s malformation of the tricuspid valve
  - Single ventricle defects
  - 20% detection rate
  - Transposition of great arteries (TGA)
  - Tetralogy of Fallot (TOF)
  - Transposition arterial
  - 10% detection rate
  - Total anomalous pulmonary venous return (TAPVR)
  - Aortic arch anomalies such as coarctation

Prenatal Diagnosis of CCHD

Normal fetal echocardiogram

Video

Courtesy of Dennis Wood, RDMS
Prenatal Diagnosis of CCHD

Normal fetal echocardiogram – Color Doppler

Video

Indications

Prenatal Congenital Heart Defects

New Jersey Critical

Indications

Normal

Maternal

Family

Nonimmune

Abnormalities

Association

Prenatal

Twin

Absent

Single

Factors

History

Factors

3rd degree

Screening

Disorders, – Rubella

– Aneuploidy

Aneuploidy

Screening

– Patent ductus arteriosus – volume overload on fetal heart

– Monochorionic twinning with

– Twin to twin transfusion syndrome (TTTS)

– Nonimmune hydrops fetalis and effusions

Indications for a directed fetal echocardiogram

Maternal Factors

– Diabetes mellitus

– Phenylketonuria

– Autoimmune disease and autoantibody positivity

– Lupus – complete heart block and cardiomyopathies

– Medication exposure

– Anticonvulsants

– Lithium – especially Ebstein’s malformation

– ACE inhibitors

– Warfarin and other Coumadin derivatives

– Retinoic acid

– NSAIDs

Indications for a directed fetal echocardiogram

Maternal Factors (continued)

– Infection – Rubella

– Assisted reproductive technology – IVF

Family History

– Maternal heart disease: 3–7% risk of CHD recurrence

– Paternal heart disease: 2–3% risk (greater if BAV/AS)

– AFFECTED SIBLINGS: 2–6% risk of CHD recurrence

– 2nd and 3rd degree relatives low risk of CHD recurrence

– Diseases, disorders or syndromes with Mendelian inheritance and associated cardiac phenotypes

– 22q11 deletion, Alagille syndrome or Williams syndrome

Indications for a directed fetal echocardiogram

Fetal Factors

– Abnormalities of umbilical cord and venous system

– Single umbilical artery – 3.9% risk of CHD

– Absent ductus venosus – volume overload on fetal heart

– Monochorionic twinning with

– Twin to twin transfusion syndrome (TTTS)

– Nonimmune hydrops fetalis and effusions


Recommended Reading

Indications for a directed fetal echocardiogram

Obstetrical screening limitations

– Remember that 90% of CHD occurs in low-risk pregnancies with none of the above noted indications for a directed fetal echocardiogram

– Even though up to 99% of women in the United States giving birth to babies with serious CHD had obstetrical ultrasounds only about 30% of the fetuses were identified prenatally

– Need for improving detection on obstetrical ultrasounds

– ‘Need for routine fetal echo in all pregnancies

Indications for a directed fetal echocardiogram


Recommended Reading

Indications for a directed fetal echocardiogram

Obstetrical screening limitations

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Recommended Reading
Prenatal Diagnosis of CCHD

**Hypoplastic left heart syndrome (HLHS)**

Four chamber view

Short axis view

www.cdc.gov

www.emedicine.medscape.com

Prenatal Diagnosis of CCHD

**Tetralogy of Fallot (TOF)**

www.cdc.gov

Prenatal Diagnosis of CCHD

**d-Transposition of the Great Arteries (d-TGA)**

LV RA LA A0

Prenatal Diagnosis of CCHD

**Total Anomalous Pulmonary Venous Return (TAPVR) and Coarctation of the Aorta**

Both may present with RV>LV size disproportion but the defects are difficult to image

Prenatal Diagnosis of CCHD

**Benefits of prenatal detection of Major CHD**

- Assess for possible chromosome abnormalities (up to 21%) or other malformations associated with major CHD
- Plan for birth and early treatment
- Site of delivery
- CHOP Special Delivery Unit
- Decreased morbidity and mortality compared to babies with unrecognized CCHD at time of delivery
- Education of parents about baby's heart defect and treatment plan in a controlled setting
- Address any psychological issues early
- Option for pregnancy termination if desired

Prenatal Diagnosis of CCHD

**Impact of prenatal diagnosis of CHD**

- Children's Hospital of NY-Presbyterian, Columbia University Medical Center
  - 2004-2009 total of 993 infants with major CHD of which 68.3% had prenatal diagnosis
  - Increased odds of scheduled delivery
  - Increased odds of induction of labor
  - No change in C-section rate
  - Increased odds of delivery at <39 weeks
  - No association with preoperative or predischarge mortality

Prenatal Diagnosis of CCHD

Future of prenatal screening for CHD
- Improved ultrasound imaging quality and techniques
- MRI imaging of the fetal heart
- Genetic basis of congenital heart defects and other abnormalities
- Cell-free fetal DNA analysis and other genetic markers of congenital heart disease

Conclusions
- Prenatal diagnosis of CCHD is the optimal time
- Present day OB ultrasound evaluation of the fetal heart detects only <50% of CCHD
- 90% of CCHD occurs in low-risk pregnancies
- OB ultrasound imaging of the fetal heart must include 4-chamber, outflow tracts, 3-vessel view and situs with identification of IVC
- Until we have better prenatal screening for CHD, we must improve newborn screening, continue POxS and always be vigilant for undiagnosed CHD in all infants