CONGENITAL HEART DEFECTS:

IMPROVING THE HEALTH OF MOTHERS AND BABIES

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I have no **relevant financial or nonfinancial relationships** in the products or services described, reviewed, evaluated or compared in this presentation.

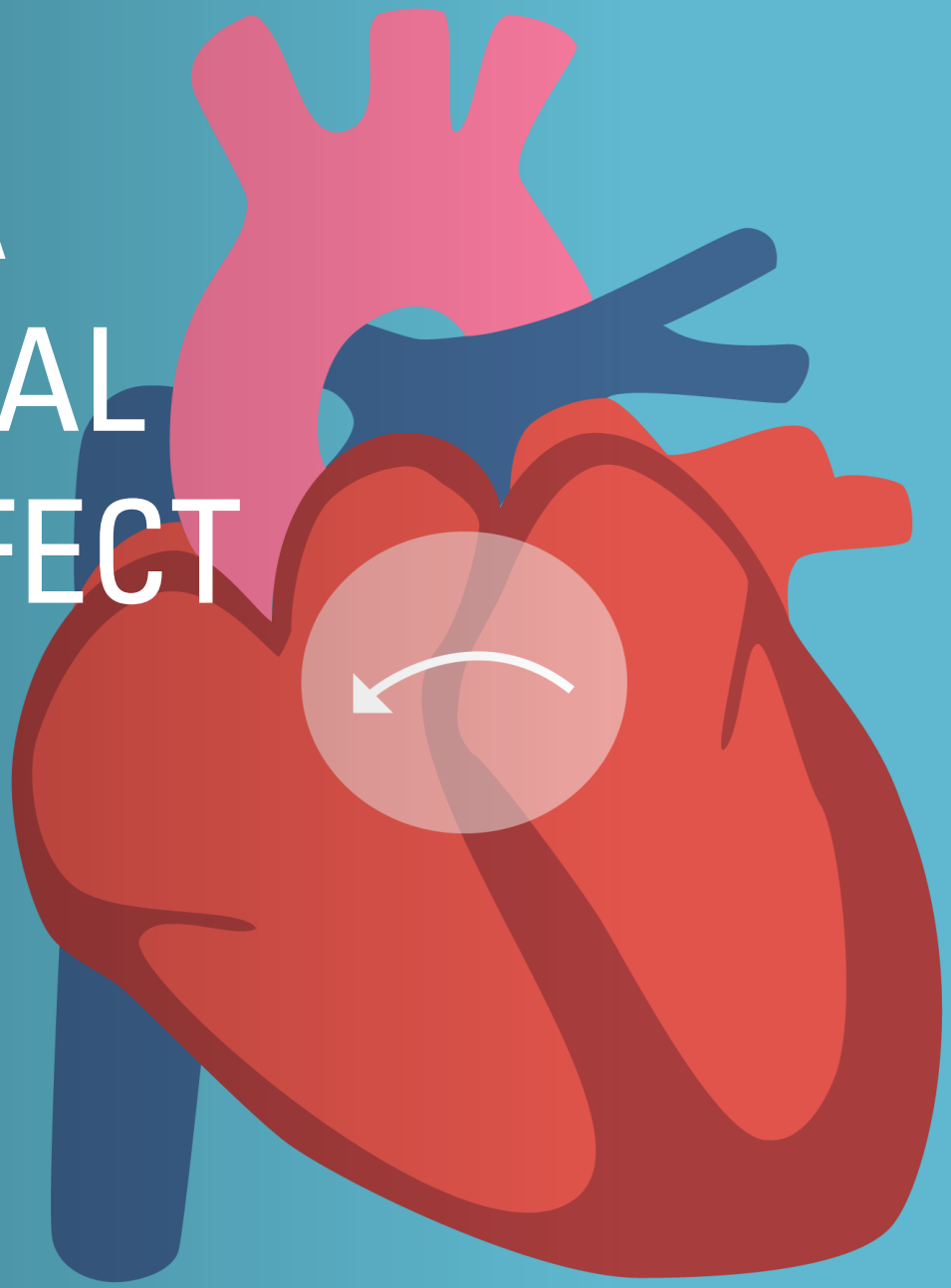
OBJECTIVES

- Describe the benefits of genetic testing during pregnancy to the mother and baby
- Describe the benefits of accurate fetal diagnosis during pregnancy of congenital heart defects to the mother and baby
- List challenges to providing optimal care to these patients

BACKGROUND

- Role as a provider in Obstetrics
- Role as a Maternal Fetal Medicine Specialist (Perinatologist)
- Multidisciplinary Approach
    1. Pediatric Cardiologist
    2. Genetics Counselor
    3. Neonatologist
    4. Geneticist
    5. Pediatric Cardiac Surgeon

WHAT IS A CONGENITAL HEART DEFECT (CHD)?



Just the basics

A. Fetal Circulation

B. Circulation after Birth

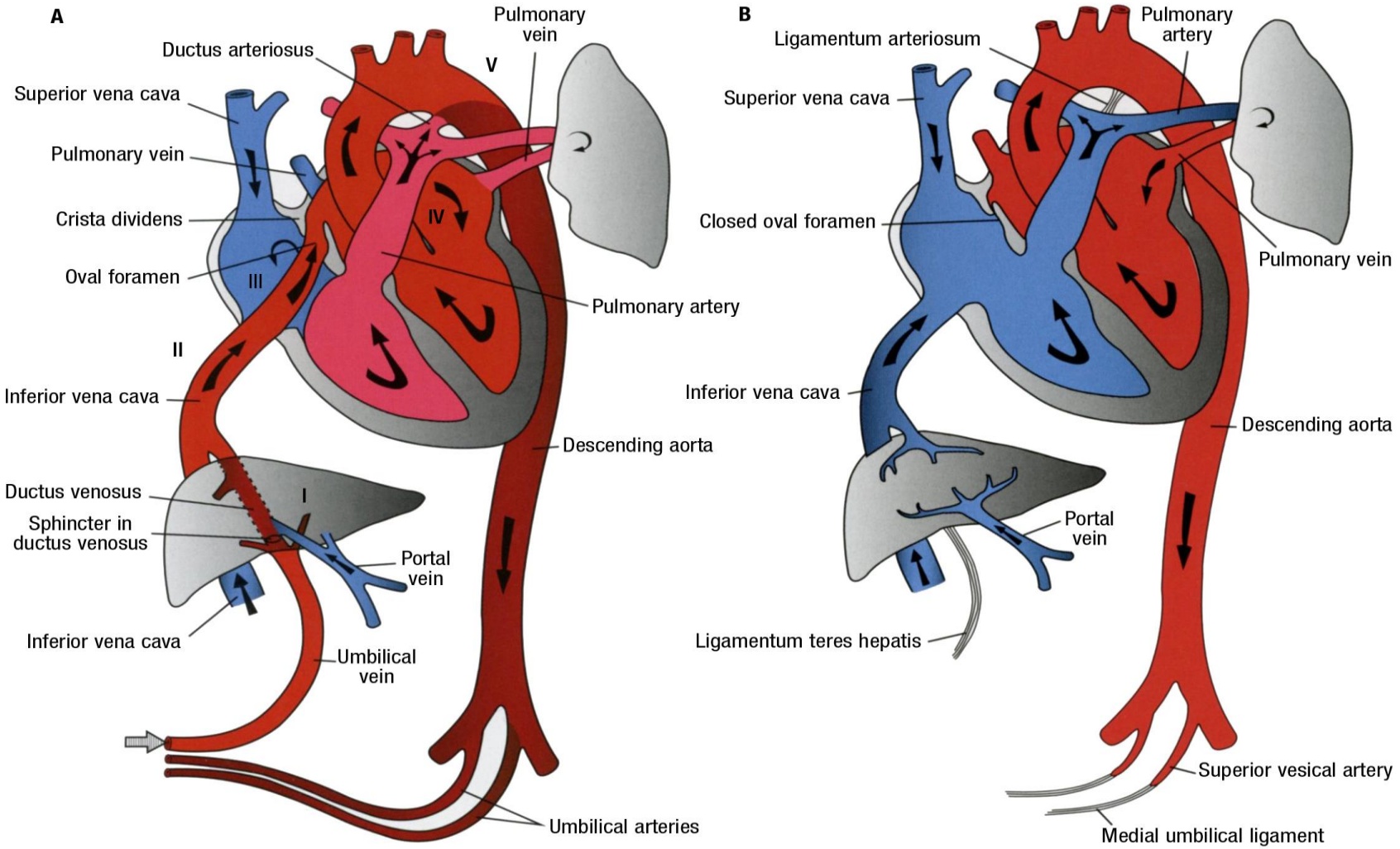


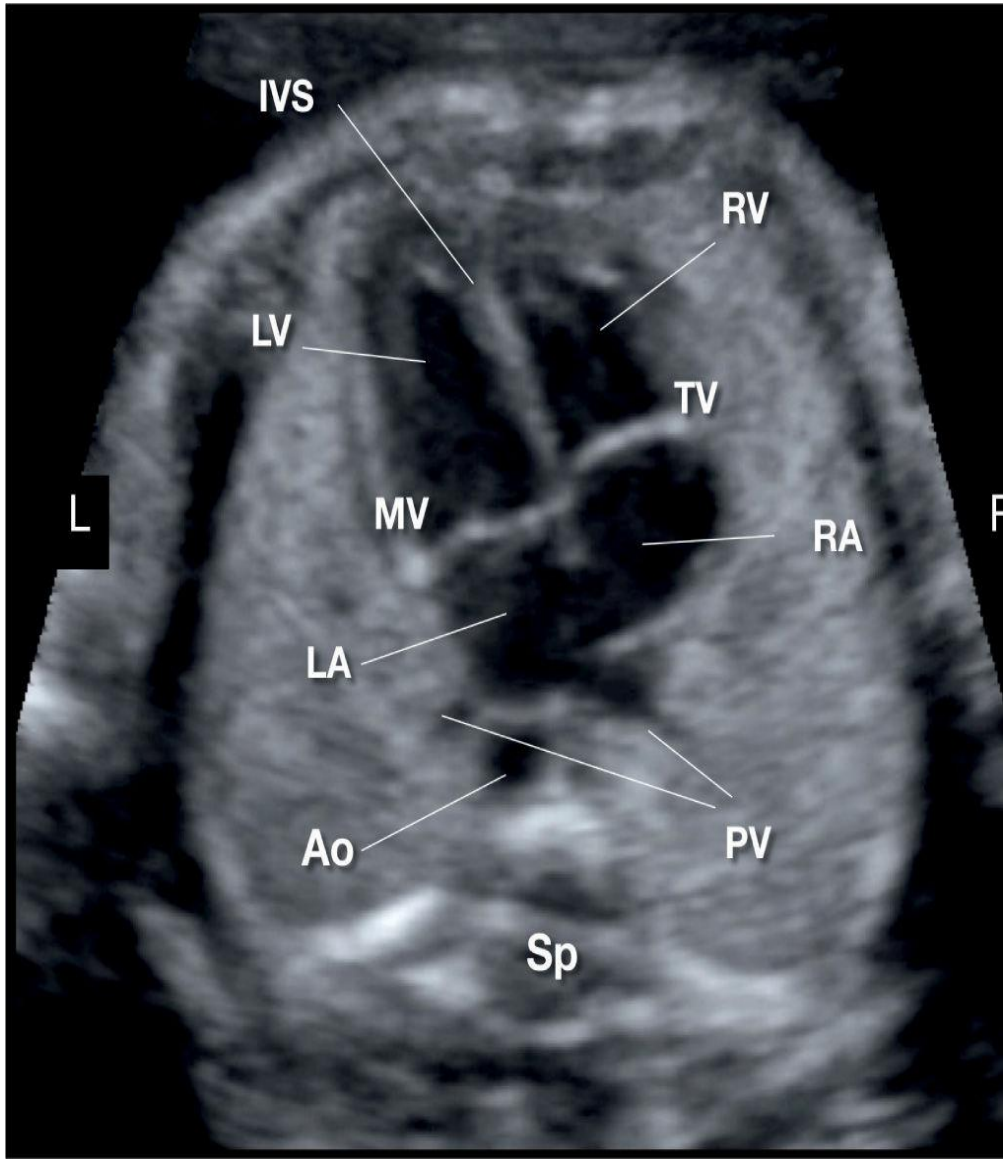
Figure 1. Normal pattern of blood flow through the heart: **A.** fetal circulation before birth; arrows indicate the direction of blood flow; oxygenated blood mixes with deoxygenated blood in the liver (I), the inferior vena cava (II), the right atrium (III), the left atrium (IV), and at the entrance of the ductus arteriosus into the descending aorta (V), **B.** human circulation after birth. (Sadler TW. Langman's medical embryology. 13th ed. Philadelphia [PA]: Wolters Kluwer; 2015.) ⇦

CONGENITAL HEART DEFECTS

- Occurs due to a disruption of the normal embryonic and postnatal development of the cardiac structures

- Categorized into cyanotic (“blue baby”) and acyanotic

- Categorization determines intervention type and timing after delivery



CONGENITAL HEART DEFECTS

Why is this important?

- Leading cause of infant death due to congenital anomalies
- In the US:
  - ✓1% of births
  - ✓4% of neonatal deaths
  - ✓30-50% of deaths due to congenital anomalies
- Most can be diagnosed prenatal

Table 1. Cyanotic and Acyanotic Congenital Heart Disease ⇦

Cyanotic Congenital Heart Disease*	Acyanotic Congenital Heart Disease
Tricuspid atresia	Atrial septal defects
Tetralogy of Fallot	Ventricular septal defects
Truncus arteriosus	Bicuspid aortic valve
Transposition of the great vessels	Coarctation of the aorta
Total anomalous pulmonary venous return	
Ebstein anomaly	
Eisenmenger syndrome	
Pulmonary atresia	
Hypoplastic left heart syndrome	

*Cyanotic congenital heart disease often is referred to as 5 Ts and 2 Es.

CASE PRESENTATION

PREGNANCY TIPS 18-20 WEEKS

The Case of the Mom with New Prenatal Care

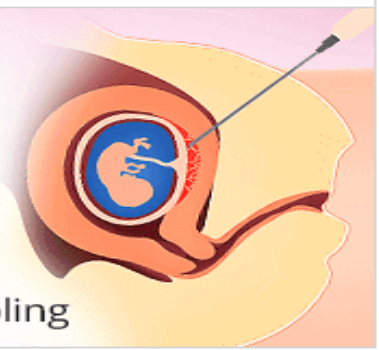
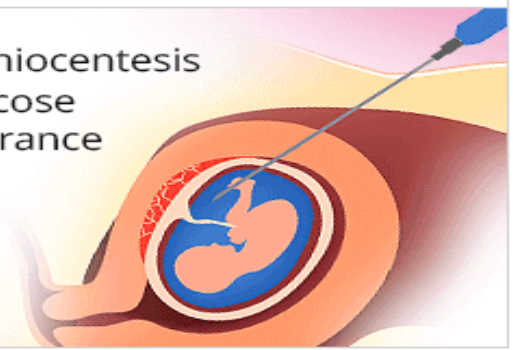
Jazelle is a 23 year old G1 P0 African American female presenting for her first OB visit. She thinks she is around 4 months pregnant but does not have regular cycles. She tells you that her mom thinks she "looks farther along". She reports being fairly healthy. This is a desired pregnancy. She is accompanied by her mom, who interjects helpful past medical history.

Upon further questioning, Jazelle's mom says she has a history of a "hole in her heart" that was found at birth. It did not cause problems and she was told that it did not require repair. Jazelle plays volleyball with no problems. Attends junior college and lives at home with her mother. She wants to become a dental hygienist. She denies any smoking, alcohol or drug use. She is in a monogamous relationship with her boyfriend of 4 years and he is excited about the baby. They plan to marry next year.

- Based on the patient's history, what should she be offered and / or ordered?
- Is there any additional information that you need?

Prenatal Testing

Prenatal testing is an array of **routine and specialized tests** are aimed at monitoring fetal development, evaluating maternal health, assessing the risk of potential complications.

	1ST TRIMESTER TESTS	2ND TRIMESTER TESTS	3RD TRIMESTER TESTS
ROUTINE	**First Prenatal Visit** Pregnancy test Blood test Urine test Dating ultrasound **1st Trimester Screening** Maternal Blood Serum Nuchal translucency	 Urine test Fetal heart rate monitoring Glucose challenge screening Quad screen Anomaly ultrasound	 Urine test Fetal heart rate monitoring Group B Strep Test Baby kick count
NON-ROUTINE	 Cell-free DNA test Chorionic villus sampling	 Amniocentesis Glucose tolerance test	 Ultrasound Non-stress test Biophysical profile Contraction stress test

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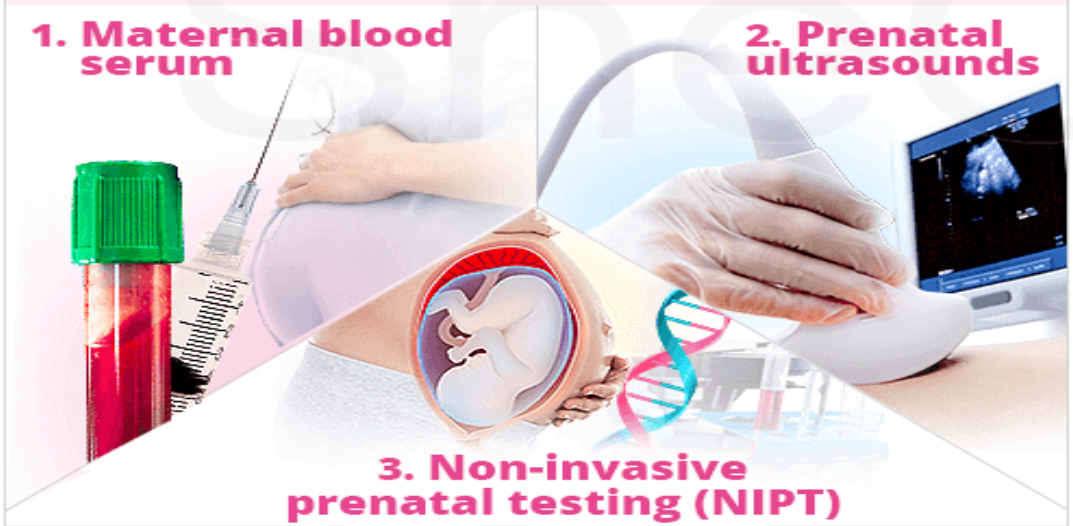
Prenatal Genetic Testing

There are **five types** of prenatal genetic tests, which can be used for screening the risk of the baby having a genetic disorder or for diagnosing such abnormalities.

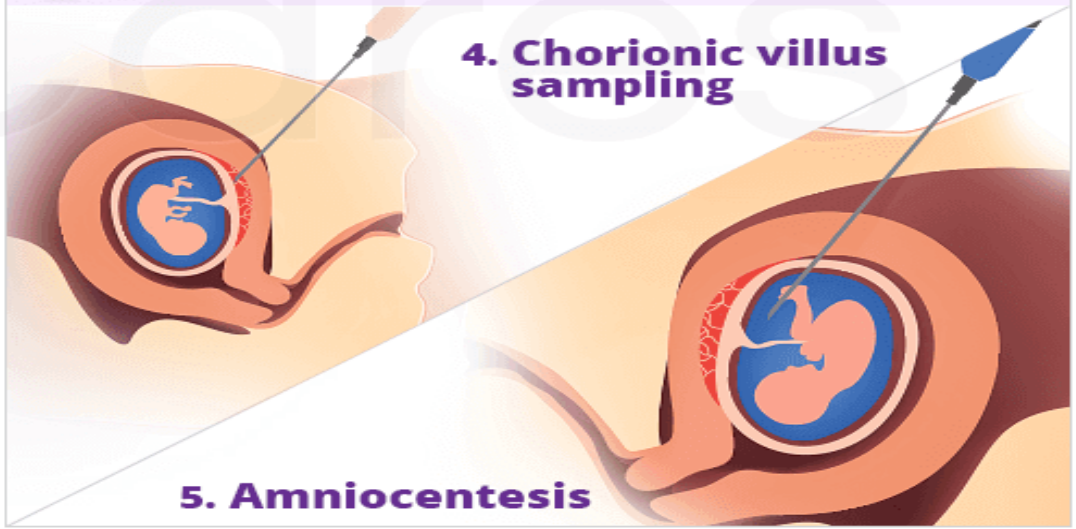
FOR SCREENING

- Evaluate the risk of genetic problems
- Non-invasive
- Offered to all women



FOR DIAGNOSING

- Confirm or rule out genetic problems
- Invasive
- Offered after abnormal screening tests or known risks factors



Benefits of Prenatal Screening and Diagnosis of CHD

- Parental counseling and preparation
- Improvement in neonatal survival
- Improvement in neonatal morbidity
- Possible opportunity for fetal treatment
- Potential harms – parental anxiety; false negative; false positive

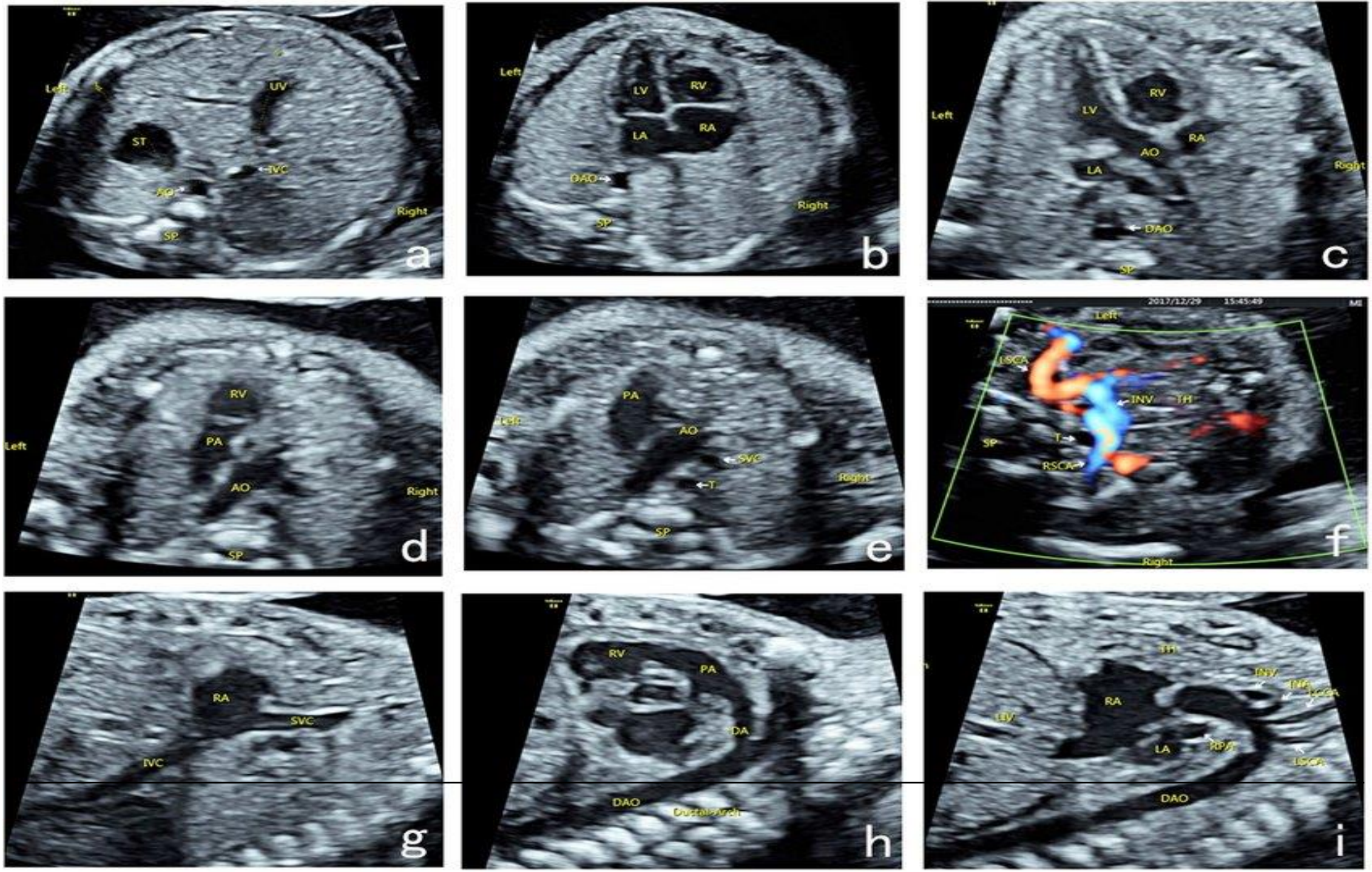
INDICATIONS FOR FETAL CARDIAC EVALUATION

Increased risk of CHD is >1% (baseline population risk is ≤ 1%). Here are some common indications of high-risk for CHD:

- Maternal pregestational diabetes mellitus
- CHD in 1st degree relative
- Suspected fetal cardiac abnormality
- Increased NT on 1st trimester screen
- Monochorionic twins

Indications for Fetal Echocardiography

Genetic	Fetal	Maternal
• First-degree relative with CHD • Mendelian syndromes that include CHD - Noonan syndrome - Tuberous sclerosis - DiGeorge syndrome • Chromosomal syndromes associated with CHD including but not limited to trisomy 21, 13 and 18 and Turner syndrome	• Abnormal ultrasound or cardiac screening - Increased NT measurement - Abnormal heart rate or rhythm - Abnormal heart views • Fetal chromosomal abnormality • Hydrops fetalis • TRAP sequence • Monochorionic twins • Extracardiac anomalies - Congenital pulmonary airway malformation – CPAM - Congenital diaphragmatic hernia – CDH - Omphalocele - Duodenal atresia - Spina bifida - Vertebral anomalies - Limb abnormalities • Abnormality of umbilical cord/placenta	• CHD • Teratogen exposure - Lithium - Antiseizure medications - Cocaine - Retinoids - Metabolic disorder - Diabetes - Phenylketonuria • Autoimmune antibodies - Systemic lupus erythematosus (SLE) • +Anti-Ro (SSA)/Anti-La (SSB) - Sjögren's • In vitro fertilization • Infections - CMV - Toxoplasmosis - Lyme disease - Etc. • BMI > 40 • Age >40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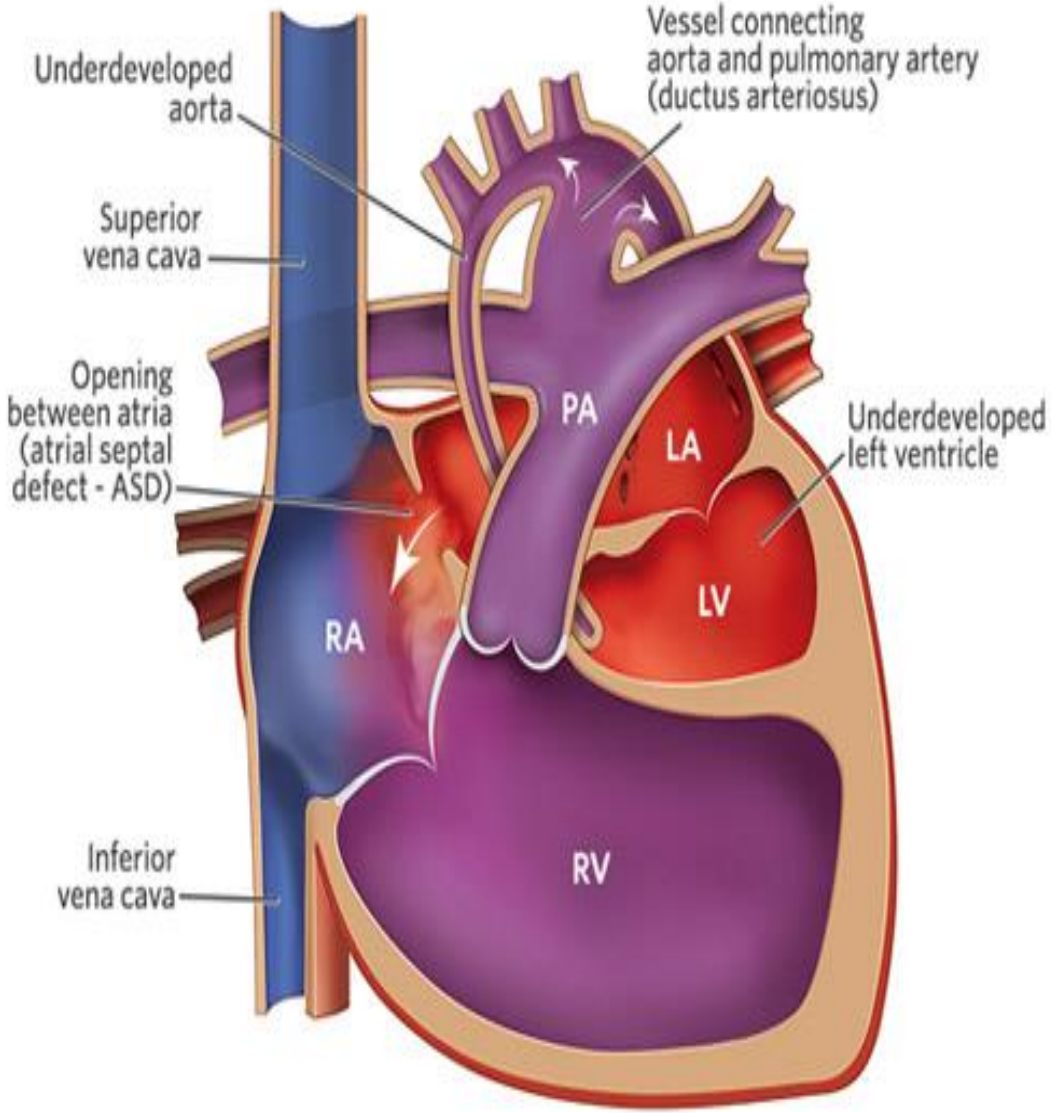
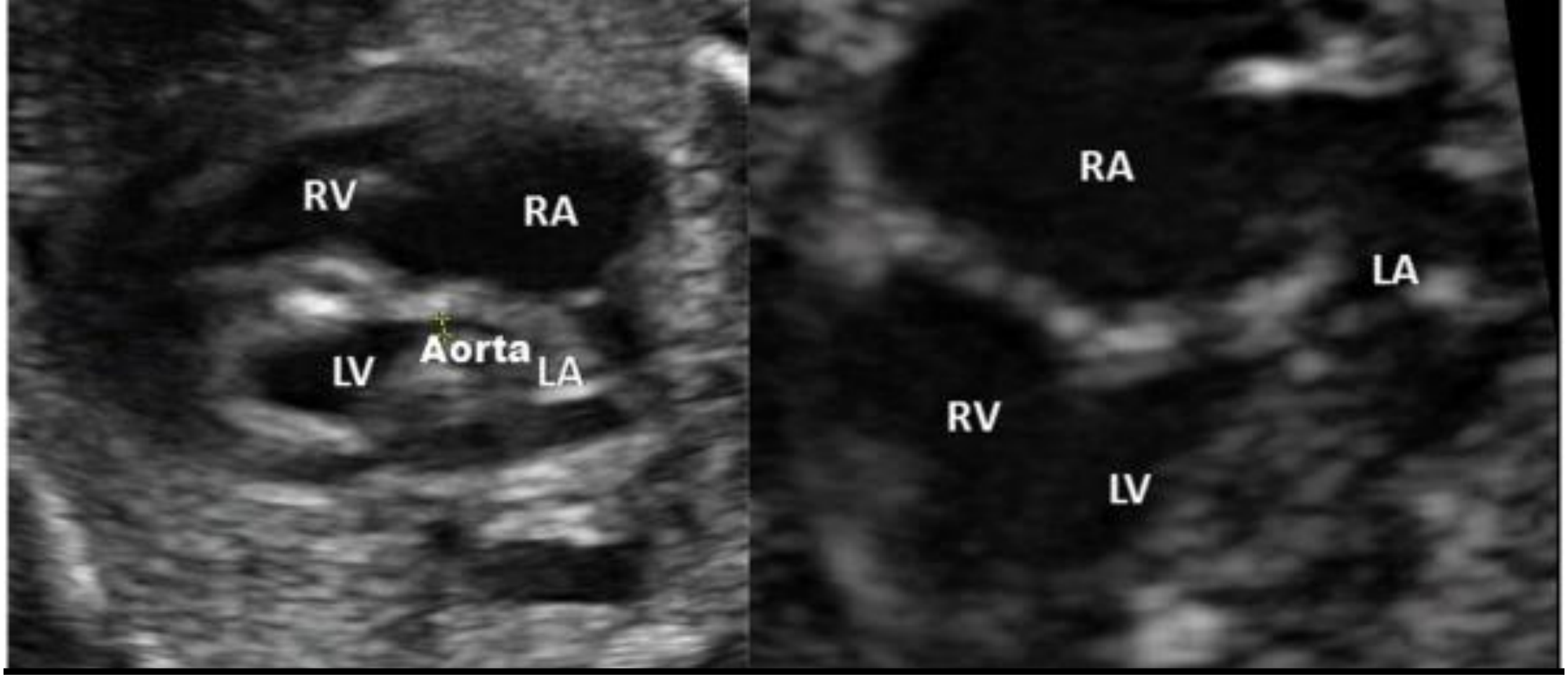
CASE CONTINUED…

- Jazelle has an ultrasound that puts her gestational age at 20 weeks 2 days
- Normal female fetus except for:
  - ➢ hypoplastic left heart (HLH) identified
- Next steps?
  - ❑Genetic consultation
  - ❑Pediatric cardiologist
  - ❑Neonatologist

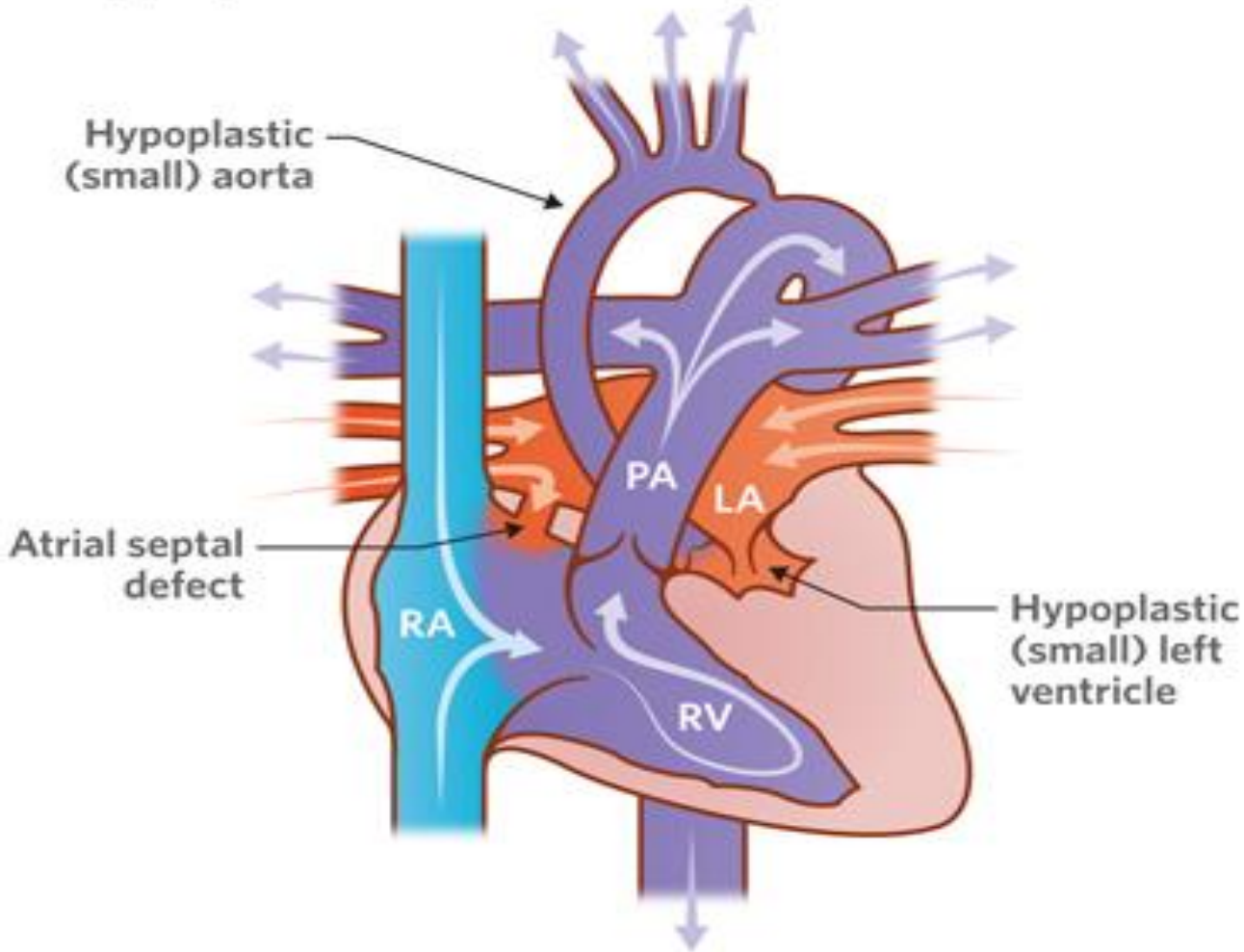


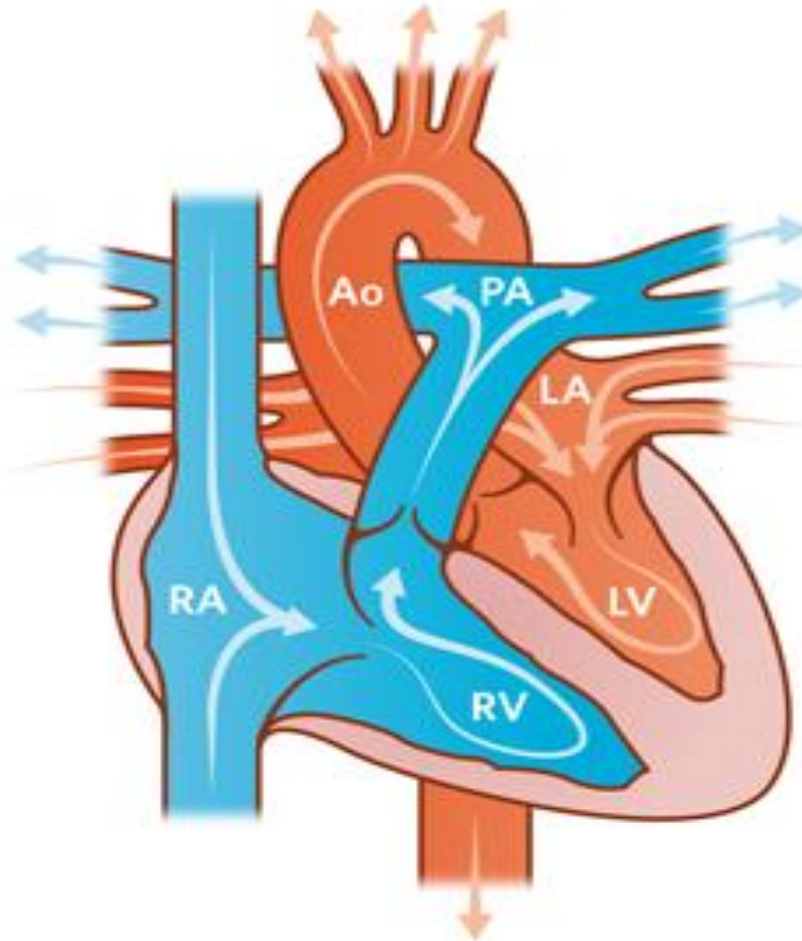


HYPOPLASTIC LEFT HEART SYNDROME (HLHS)

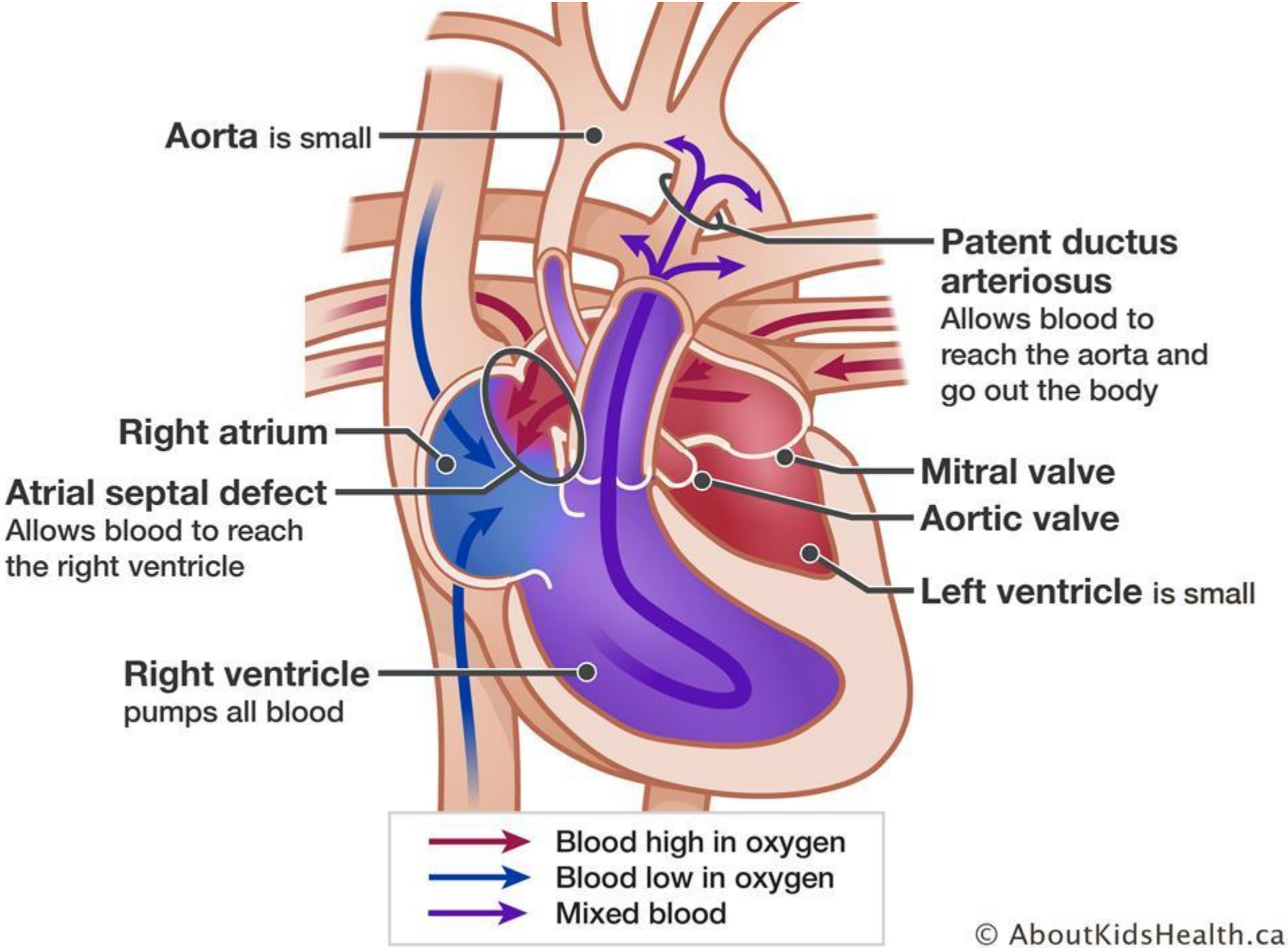
What does this mean for the mom?

Hypoplastic left heart syndrome





Normal heart and circulation


Aorta is small
Patent ductus arteriosus
Allows blood to reach the aorta and go out the body
Right atrium
Mitral valve
Atrial septal defect
Allows blood to reach the right ventricle
Aortic valve
Left ventricle is small
Right ventricle
pumps all blood
Blood high in oxygen
Blood low in oxygen
Mixed blood
© AboutKidsHealth.ca

HLHS: QUICK OVERVIEW

Terminology

- Hypoplasia of left ventricle associated with:

1. Mitral stenosis / atresia
2. Aortic stenosis / atresia
3. Hypoplastic ascending aorta and coarctation

Imaging

- LV small or non-existent
- RV is dilated with good function
- Interatrial septum bowed left to right showing direction of blood flow
- Ascending aorta / transverse arch are very small
- Retrograde filling of aortic arch = ductal dependence

HLHS: QUICK OVERVIEW

Clinical Issues

- Cause may be multifactorial; 12% prevalence of cardiac abnormalities in 1st degree relative of HLHS patients
- Male predominance of 55-67%
- Lethal in days/weeks if untreated
- If pregnancy continues, options include:

1. Comfort care
2. Surgical intervention
3. Hybrid procedure

Prognosis

- Improving surgical techniques → increased survival
- >85% success of 1st stage Norwood in certain centers
- ~100% success for Glenn and Fontan (2nd and 3rd stage) surgeries
- 6-year survival with current techniques – 64%
- RR:
    1. 2% w/ 1 sibling, 6% w/ 2
    2. 25% if autosomal recessive

Hopefully diagnosed prenatally but just in case it is missed, these are the post-delivery warning signs of HLHS in a newborn!

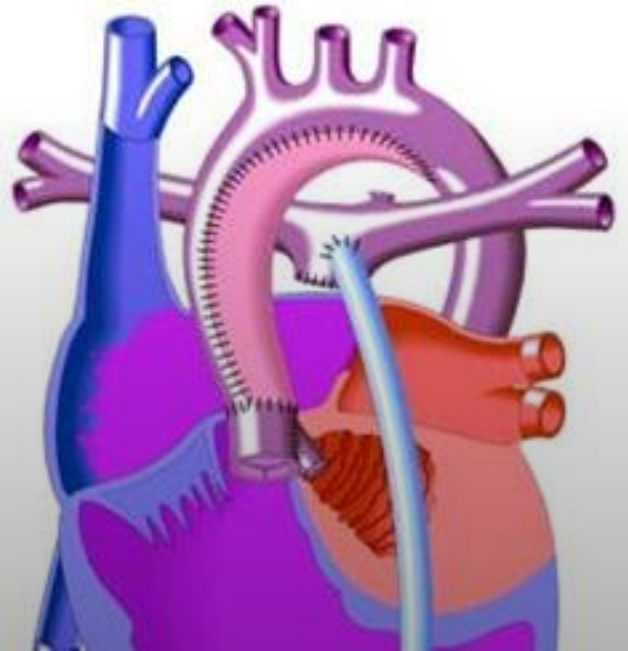
CASE: RETURN TO JAZELLE

1) Genetics: offer karyotype
2) Prenatal consultation w/ PEDS Cardiology and NICU
3) Termination (if previable is an option)
4) Options if continues pregnancy:
    a. Comfort care – no monitoring, deliver at home institution
    b. Surgical intervention – planned delivery at tertiary center specializing in cardiac care
    c. Hybrid approach – planned delivery at tertiary center specializing in cardiac care
    d. Heart transplantation – reserved mostly for surgical failures; not offered at birth
5) Other prenatal interventions: balloon valvuloplasty (severe aortic stenosis)
6) Post-delivery: the surgical route

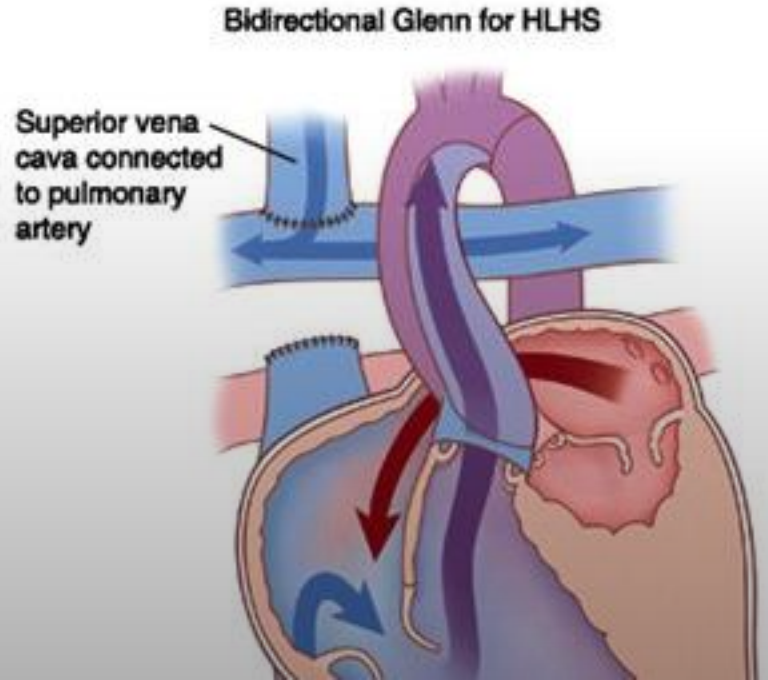
HLHS (Hypoplastic Left Heart Syndrome)

- **Norwood**
- **Bidirectional Glenn**
- **Fontan**

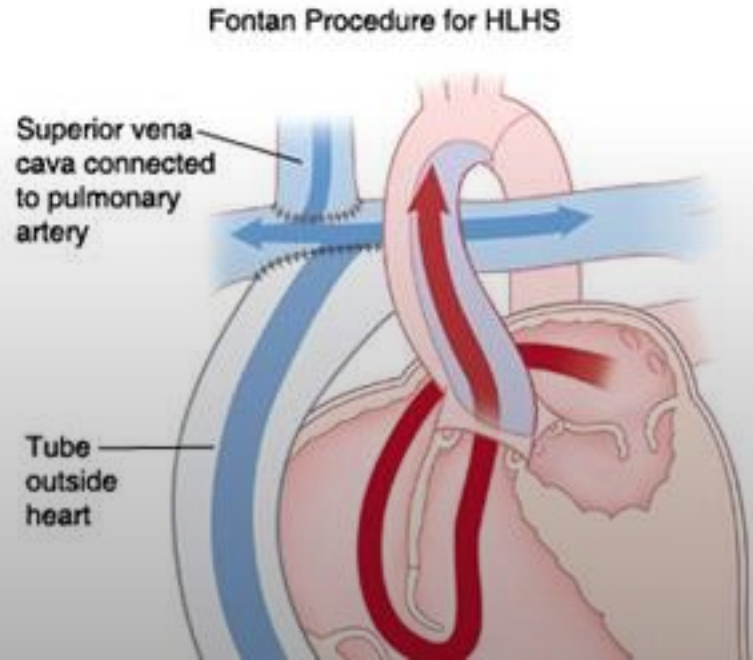
1st Repair



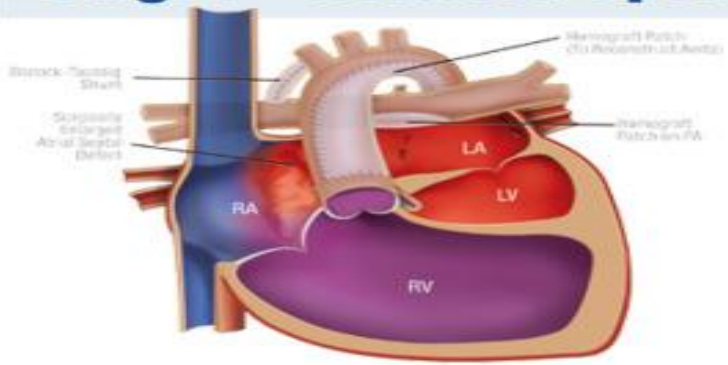
2nd Repair



3rd Repair



Stage I – Norwood Operation

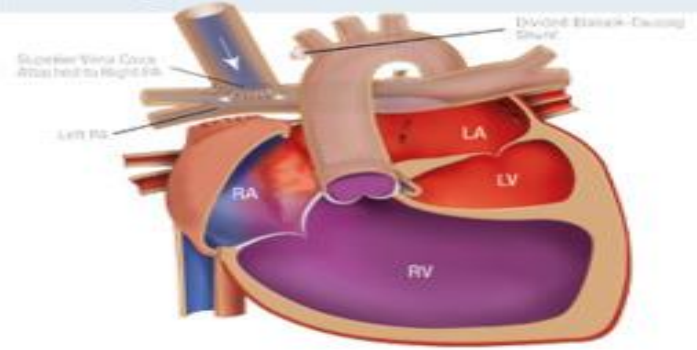


Stage I operation, also known as the Norwood procedure, occurs within a few days of birth.

- A tube is put in to connect a main branch of the pulmonary artery to the aorta to give blood flow to the lungs. The Blalock-Taussig shunt (a tube) is placed from the aorta to the pulmonary artery to give blood flow to the lungs.
- The main pulmonary artery and the small aorta are joined with a patch to make a larger aorta.
- The opening between the two top chambers (atrial septal defect) is enlarged.

** Alternatives to the Blalock-Taussig shunt may be used based on a child's individual anatomy.*

Stage II – Bidirectional Glenn Operation

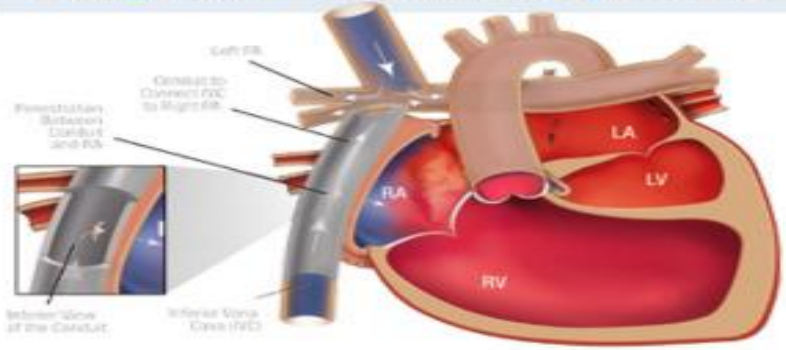


Stage II operation, also known as the Bidirectional Glenn or the Hemi-Fontan, typically occurs within four to six months of birth.

- The shunt from the Stage I Norwood operation is removed.
- The vein (superior vena cava) that carries deoxygenated (blue) blood from the upper body to the heart is redirected. It is taken off the heart, and then connected to the pulmonary arteries, so the blood flows to the lungs without passing through the heart.

** Alternatives to the bidirectional Glenn may be used based on a child's individual anatomy.*

Stage III – Fontan Operation



Stage III operation, also known as the Fontan procedure, typically occurs between one-and-a-half to four years of age.

- The vein (inferior vena cava) that carries deoxygenated (blue) blood from the lower body to the heart is redirected. A conduit (tube) is used to connect blood flow from the lower body directly to the pulmonary arteries, so the blood flows to the lungs without passing through the heart.
- A small hole (fenestration) is sometimes created between the conduit and the right atrium.

** This illustration shows Stage III of reconstruction using a technique called an extracardiac Fontan. In some children a different modification, called a lateral tunnel fenestrated Fontan, is used.*

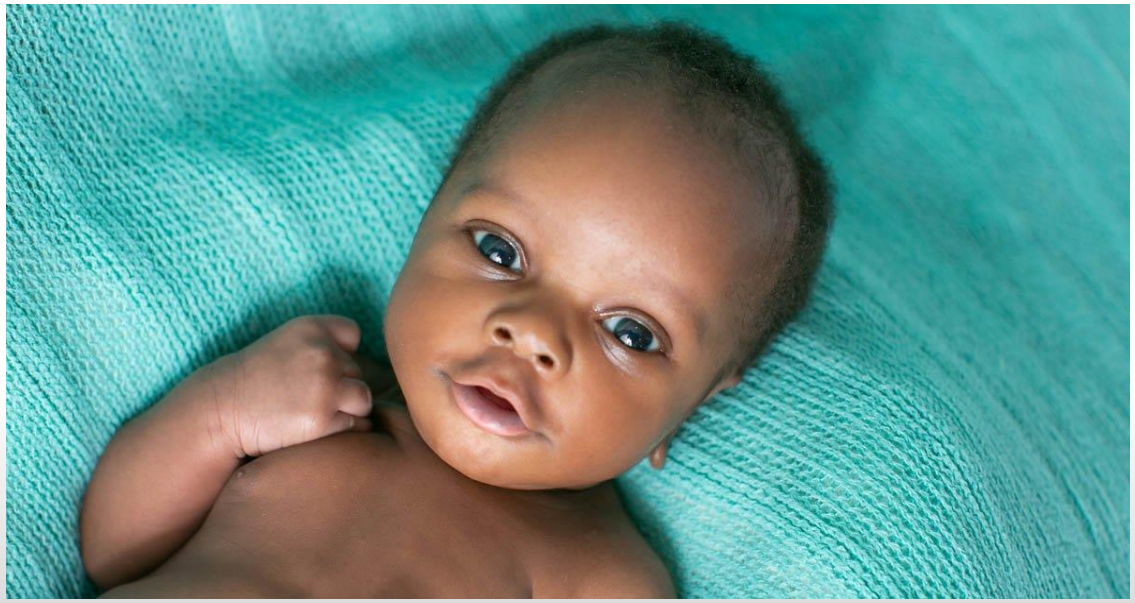
Oxygen-rich Blood Oxygen-poor Blood Mixed Blood

AO: Aorta **LA:** Left Atrium **LV:** Left Ventricle **PA:** Pulmonary Artery **RA:** Right Atrium **RV:** Right Ventricle

Case: Jazelle's BABY—Ophelia, Follow-up

1) Long-term risk: dependent on options chosen (e.g. surgery)

2) Long-term follow-up for baby girl Ophelia:
   - Lifelong follow-up with a cardiologist
   - Annual clinical evaluation
   - Regular laboratory testing
   - Annual EKG
   - Annual CXR
   - Stress testing
   - Endocarditis prophylaxis
   - Long-term anticoagulation



Case: Jazelle's BABY—Ophelia, Follow-up

3) Morbidities in children with HLHS after Fontan procedure
    a. Exercise intolerance
    b. Arrhythmia
    c. Thromboembolic disease (e.g. PE, stroke)
    d. Protein-losing enteropathy
    e. Neurocognitive disabilities (e.g. learning disabilities, ADD/ADHD)

4) 30-year survival rate s/p Fontan procedure ~85%

REPRODUCTIVE RISK FOR OPHELIA

PRECONCEPTION

- Discouraged if poor functional capacity, h/o heart failure or ventricular function < 40%, arrhythmias, enteropathy, intracardiac thrombi, or cyanosis (RA sat <90%)
- Any pregnancy in woman with HLHS is HIGH RISK. Complications include:
    a. Arrhythmias
    b. Thrombotic / bleeding events
    c. SAB, PTB, FGR and PPH
    d. Recurrence risk for Ophelia's baby

CONCLUSIONS

We can do better!

For fetal conditions like HLHS prenatal diagnosis with fetal ECHO confirms 95% of the time.

KEY ➔

prompt patients to start prenatal care early, screen appropriately and refer if indicated


The way to get started
is to quit talking and
begin doing.
Walt Disney

OBJECTIVES

- Describe the benefits of genetic testing during pregnancy to the mother and baby
- Describe the benefits of accurate fetal diagnosis during pregnancy of congenital heart defects to the mother and baby
- List challenges to providing optimal care to these patients

BETTER OBJECTIVES

- Become better at screening the fetal heart for CHD
- Become better at diagnosing and evaluating common forms of fetal cardiac malformations
- Understand the basic approach to fetal and neonatal management of fetal cardiac malformations
- Understand the prognosis associated with the most common forms of fetal heart disease (example, HLHS)

References

All images and videos were obtained to the best of my knowledge from public domain on the internet and I make no claims to own them or to being the creator of the above stated material.

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THANK YOU

Any questions?