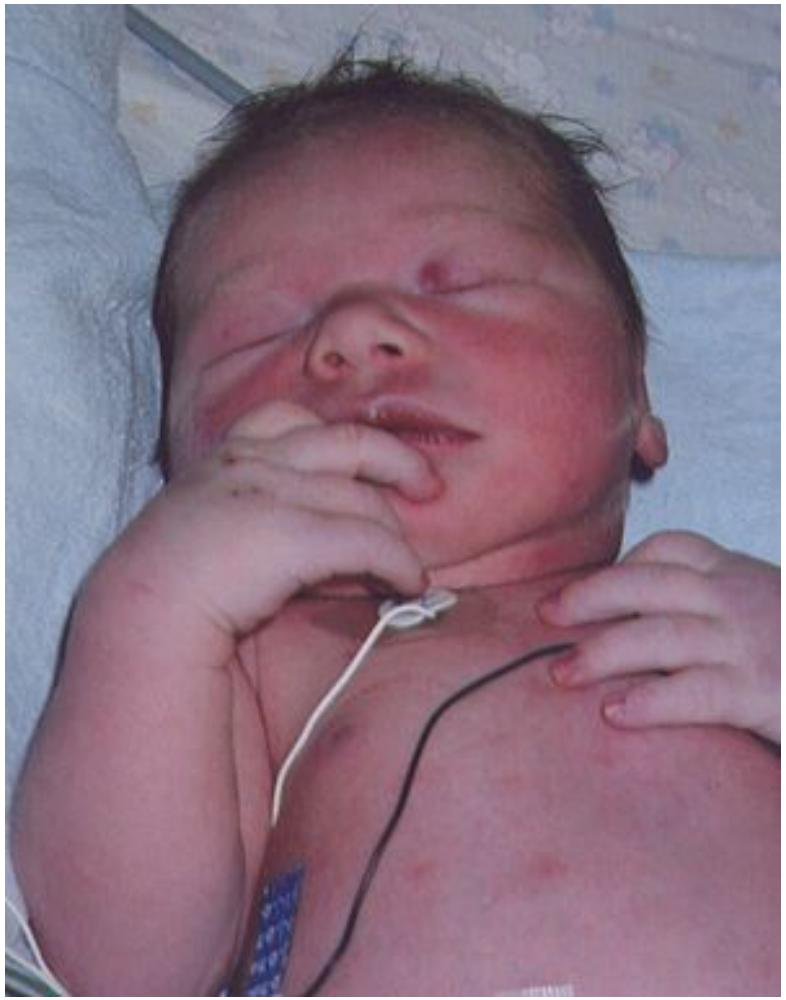
# MEET BRIAN

# Brian's Story

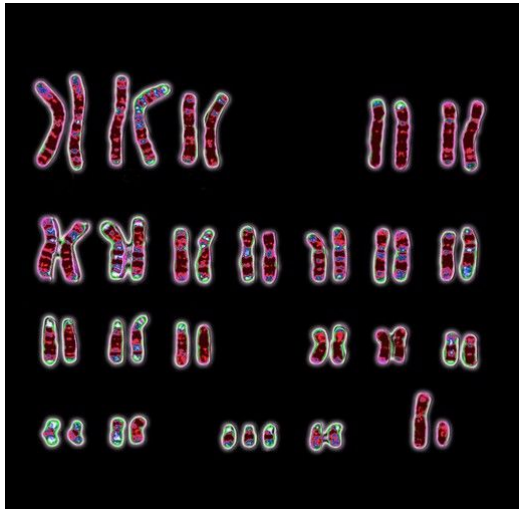
- **Born 1989 – affluent parents in the early forties**
- **Diagnosed with Down syndrome at birth**
- **Stayed at home with around the clock hired help**
- **Parents who were devastated and ashamed**
- **Older brother away at boarding school**

# Brian's Health

- 1989 – diagnosed with Trisomy 21, 24 hours after birth
- Blue-tinged skin, heart murmur
- Diagnosed with Tetralogy of Fallot
- Temporary surgery for bypass shunt
- 6 months later Intracardiac surgery
- Social isolation
- Around the clock care



# Down Syndrome Overview



- There are 23 pairs of chromosomes, for a total of 46.
- Half the chromosomes come from the egg, and the other half from the sperm.
- This XY chromosome pair includes the X chromosome from the egg and the Y chromosome from the sperm.
- In Down's syndrome, there is an additional copy of chromosome 21, resulting in three copies instead of the normal two copies.
- 
- Trisomy 21 (also known by the karyotype 47,XX,+21 for females, 47,XY,+21 for males) is caused by a failure of the 21$^{st}$ chromosome to separate during egg and sperm development (nondisjunction).

https://www.mayoclinic.org/diseases-conditions/down-syndrome/symptoms-causes/syc-20355977

# Three Genetic Variations

- **Trisomy 21.** About 95 percent of the time, Down syndrome is caused by trisomy 21 — the person has three copies of chromosome 21, instead of the usual two copies, in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.
- **Mosaic Down syndrome.** In this rare form of Down syndrome, a person has only some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.
- **Translocation Down syndrome.** Down syndrome can also occur when a portion of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional genetic material from chromosome 21 attached to another chromosome.



https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html

# Risk Factors

- **Advancing maternal age.** A woman's chances of giving birth to a child with Down syndrome increase with age because older eggs have a greater risk of improper chromosome division. A woman's risk of conceiving a child with Down syndrome increases after 35 years of age. However, most children with Down syndrome are born to women under age 35 because younger women have far more babies.

- **Being carriers of the genetic translocation for Down syndrome.** Both men and women can pass the genetic translocation for Down syndrome on to their children.

- **Having had one child with Down syndrome.** Parents who have one child with Down syndrome and parents who have a translocation themselves are at an increased risk of having another child with Down syndrome. A genetic counselor can help parents assess the risk of having a second child with Down syndrome.

# Screening During Pregnancy

- ***First trimester*** **combined two step test:**
- ***Blood test*****-measures levels of pregnancy-associated plasma protein-A (PAPP-A) and human chorionic gonadotropin (HCG). Abnormal levels of both may indicate a problem with the baby.**
- ***Nuchal translucency test*****-an ultrasound is used to measure a specific area on the back of the fetus's neck. When abnormalities are present, more fluid tends to collect in this neck tissue.**
- ***Second trimester:***
- ***Blood test*****-measures four pregnancy-associated substances: alpha fetoprotein, estriol, HCG, and inhibin A.**
- **If screening test results are positive or you are at high risk, more testing to confirm the diagnosis may be done.**
- ***Chorionic villus sampling (CVS)*****-cells are taken from the placenta and analyzed. Usually done in the 1st trimester. The risk of miscarriage is very low.**
- ***Amniocentesis*****-sample of amniotic fluid is withdrawn using a needle inserted into the uterus to analyze the fetus chromosomes. Done in 2nd trimester, risk of miscarriage is very low.**

https://www.mayoclinic.org/diseases-conditions/down-syndrome/diagnosis-treatment/drc-20355983

# Common Features of Down Syndrome

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet
- Excessive flexibility
- Tiny white spots on the colored part (iris) of the eye called Brushfield's spots
- Short height

https://www.mayoclinic.org/diseases-conditions/down-syndrome/symptoms-causes/syc-20355977

# Other Common Features

- **May be of average size, but typically grow slowly and remain shorter than other children their age**
- **Most children with Down Syndrome have mild to moderate cognitive impairment**
- **Language is delayed**
- **Short- and long-term memory is affected**


Vanellus Foto

https://www.mayoclinic.org/diseases-conditions/down-syndrome/symptoms-causes/syc-20355977

# Complications

- **Heart defects.** About half the children with Down syndrome are born with some type of congenital heart defect. These heart problems can be life-threatening and may require surgery in early infancy.
- Gastrointestinal (GI) defects. GI abnormalities occur in some children with Down syndrome and may include abnormalities of the intestines, esophagus, trachea and anus. The risk of developing digestive problems, such as GI blockage, heartburn (gastroesophageal reflux) or celiac disease, may be increased.
- **Immune disorders.** Because of abnormalities in their immune systems, people with Down syndrome are at increased risk of developing autoimmune disorders, some forms of cancer, and infectious diseases, such as pneumonia.
- **Sleep apnea.** Because of soft tissue and skeletal changes that lead to the obstruction of their airways, children and adults with Down syndrome are at greater risk of obstructive sleep apnea.
- **Obesity.** People with Down syndrome have a greater tendency to be obese compared with the general population.
- Spinal problems. Some people with Down syndrome may have a misalignment of the top two vertebrae in the neck (atlantoaxial instability). This condition puts them at risk of serious injury to the spinal cord from overextension of the neck.
- **Leukemia.** Young children with Down syndrome have an increased risk of leukemia.
- **Dementia.** People with Down syndrome have a greatly increased risk of dementia — signs and symptoms may begin around age 50. Having Down syndrome also increases the risk of developing Alzheimer's disease.
- **Other problems.** Down syndrome may also be associated with other health conditions, including endocrine problems, dental problems, seizures, ear infections, and hearing and vision problems.

# Coping & Support

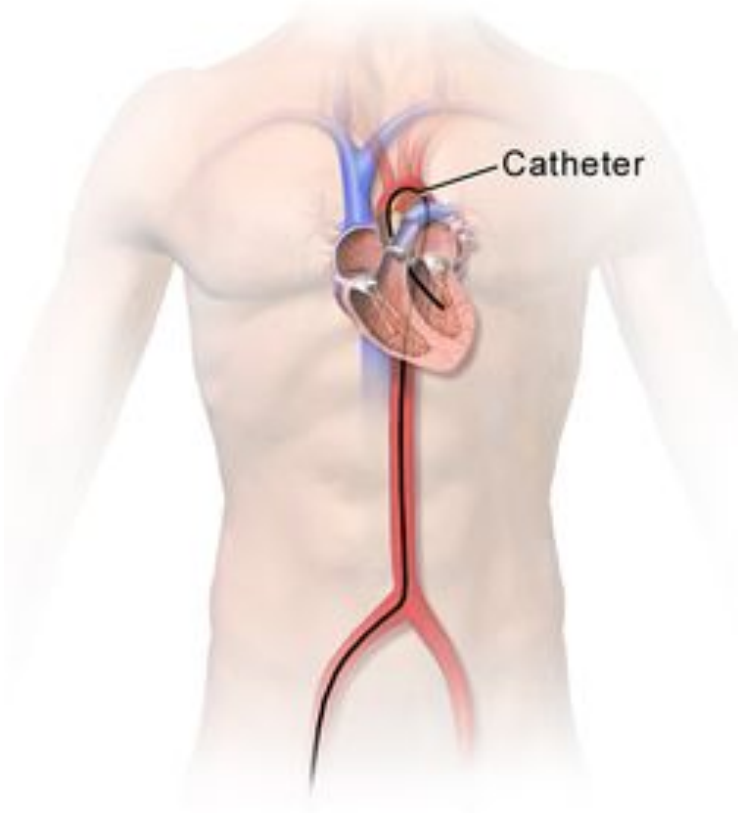
- Parents may experience a range of emotions, including anger, fear, confused, scared, worry, and sorrow.
- The best antidote for fear and worry is information and support.
- Ask your health care provider about early intervention programs
- Learn about educational options for school
- Seek out other families who are dealing with the same issues
- Participate in social and leisure activities
- Encourage independence
- Prepare for the transition to adulthood
- Most people with Down syndrome live full lives. Go to mainstream schools, participate in the community and have jobs.

https://www.mayoclinic.org/diseases-conditions/down-syndrome/diagnosis-treatment/drc-20355983

# Initial Diagnostic Tests

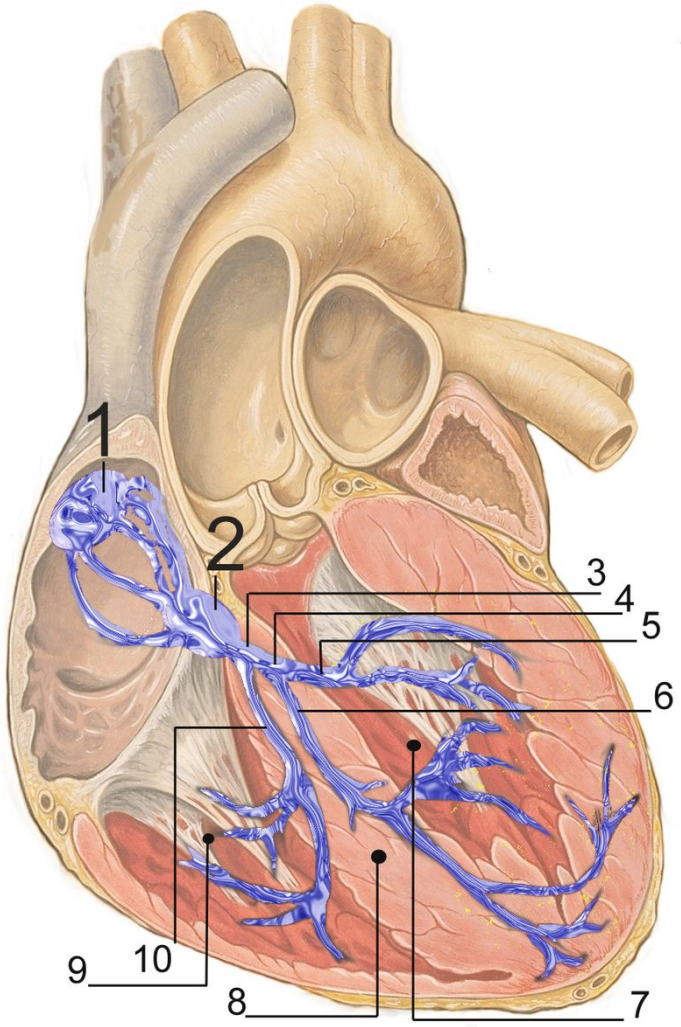
- **Chromosomal karyotype (blood test)-to diagnosis congenital abnormalities**
- **Echocardiogram-uses sound waves to produce moving images, providing information on structure and function of the heart**
- **Electrocardiogram-detects cardiac abnormalities by measuring the electrical activity generated by the heart**
- **Chest x-ray-can show complications related to lung and heart problems**
- **Oxygen level measurement-estimates the amount of oxygen in your blood**
- **Cardiac catheterization-used to evaluate heart function and diagnose cardiovascular conditions**

# Heart Catheterization



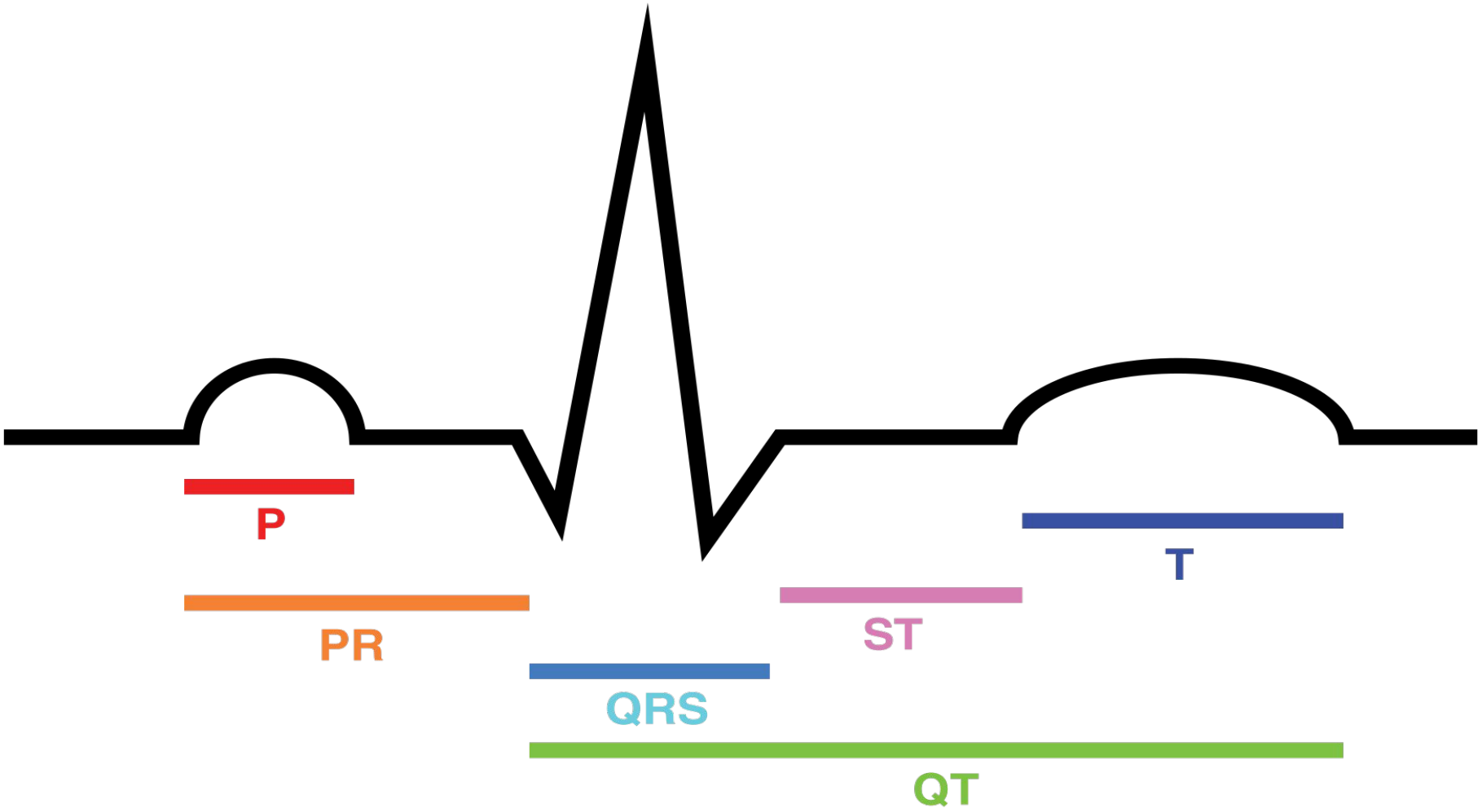

Left Heart Catheter

# Electrical Conduction System of the Heart



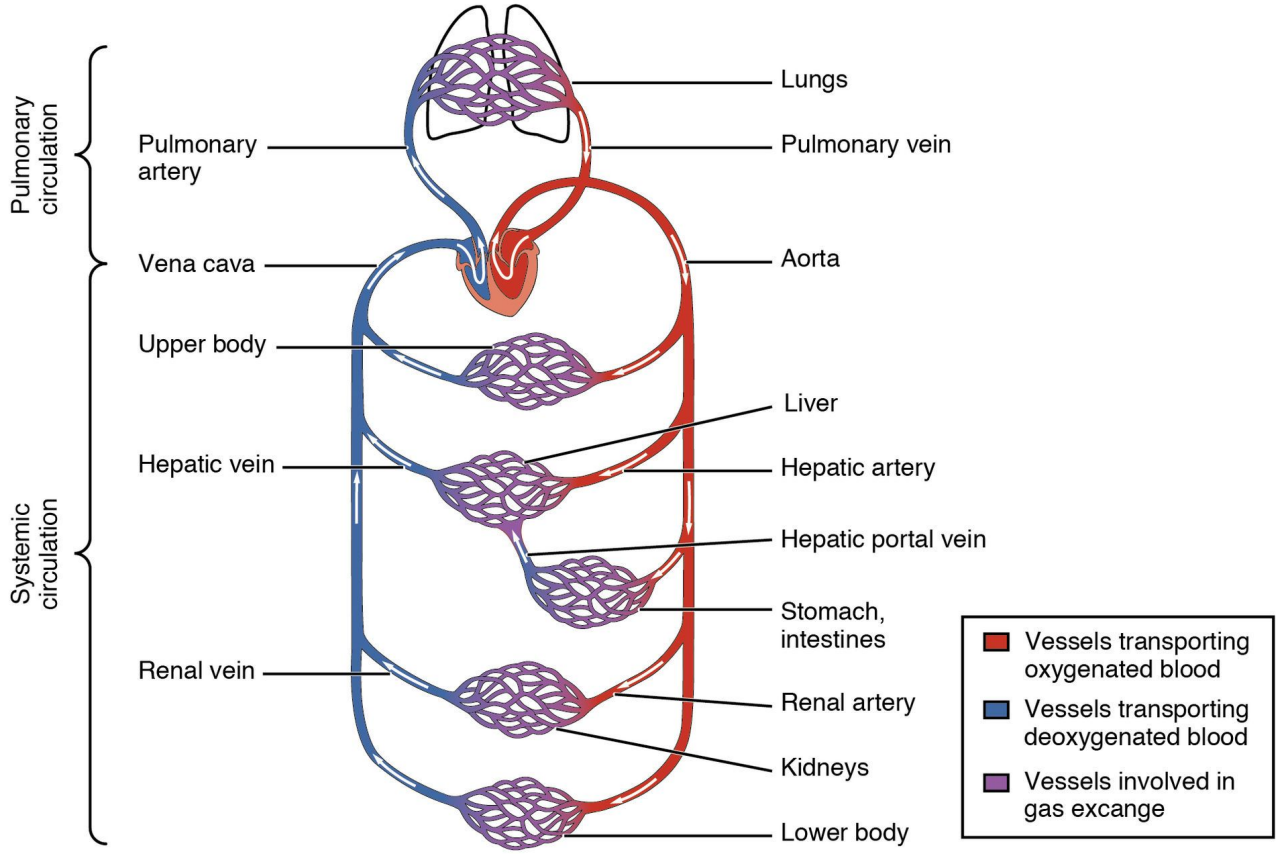

1. Sinoatrial node
2. Atrioventricular node
3. Bundle of His
4. Left bundle branch
5. Left posterior fascicle
6. Left anterior fascicle
7. Left ventricle
8. Ventricular septum
9. Right ventricle
10. Right bundle branch

# Electrocardiogram Complex



# Chambers & Circulation through the Heart

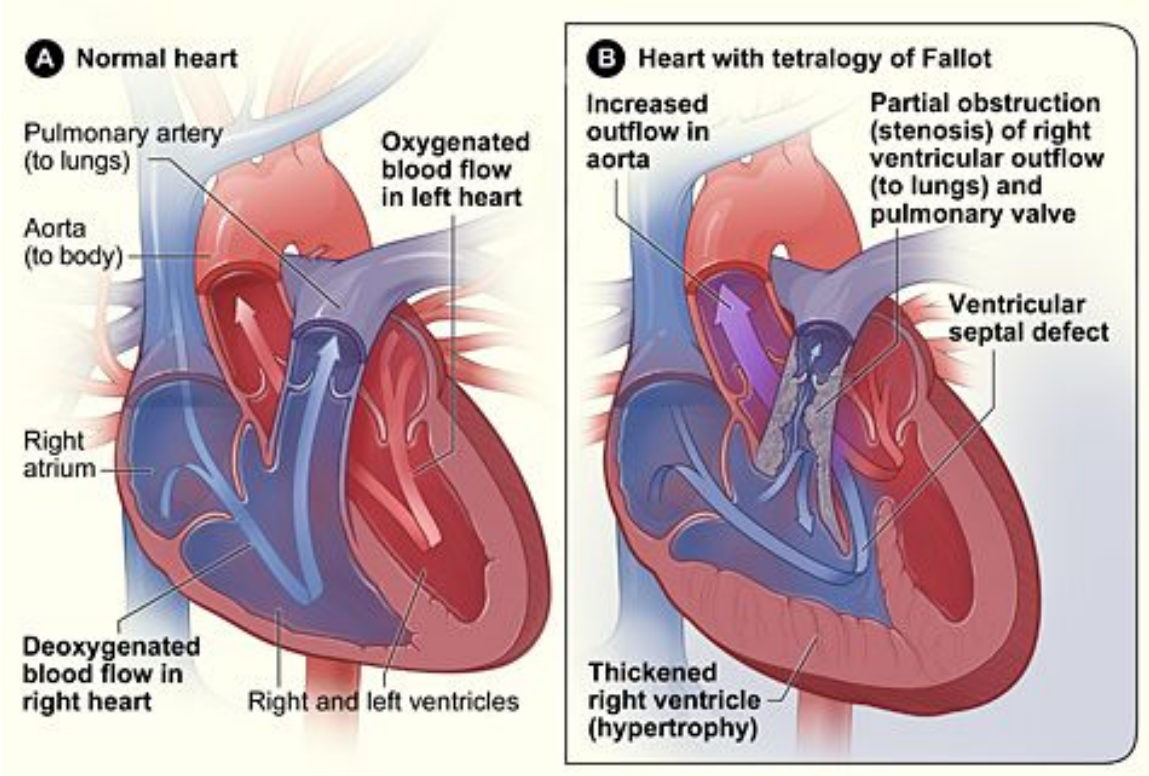
- **Blood flows from the right atrium to the right ventricle, where it is pumped into the pulmonary circuit.**
- **The blood in the pulmonary artery branches is low in oxygen but relatively high in carbon dioxide.**
- **Gas exchange occurs in the pulmonary capillaries (oxygen into the blood, carbon dioxide out), and blood high in oxygen and low in carbon dioxide is returned to the left atrium.**
- **From here, blood enters the left ventricle, which pumps it into the systemic circuit.**
- **Following exchange in the systemic capillaries (oxygen and nutrients out of the capillaries and carbon dioxide and wastes in), blood returns to the right atrium and the cycle is repeated.**




https://openstax.org/books/anatomy-and-physiology/pages/19-1-heart-anatomy?query=heart

# Facts About Tetralogy of Fallot

- **Birth defect that affects normal blood flow through the heart.**
- **Happens when the heart does not form correctly as the baby grows & develops in the womb.**
- **Made up of 4 defects of the heart and its blood vessels.**



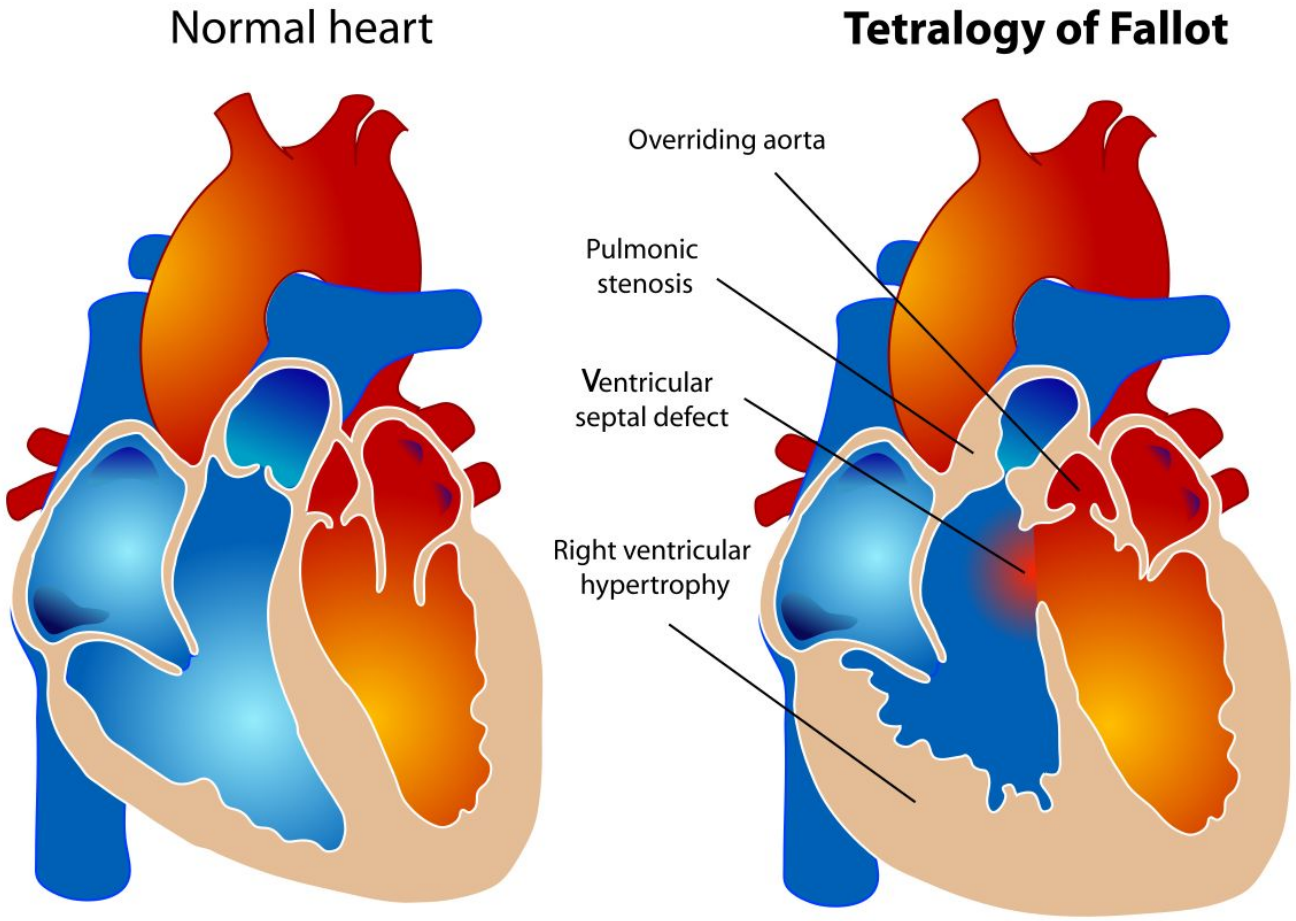

https://commons.wikimedia.org/wiki/File:Tetralogy_fallot.jpg

# The 4 Abnormalities:

- **Pulmonary valve stenosis.** Pulmonary valve stenosis is a narrowing of the pulmonary valve — the valve that separates the lower right chamber of the heart (right ventricle) from the main blood vessel leading to the lungs (pulmonary artery).
- Narrowing (constriction) of the pulmonary valve reduces blood flow to the lungs. The narrowing might also affect the muscle beneath the pulmonary valve. In some severe cases, the pulmonary valve doesn't form properly (pulmonary atresia) and causes reduced blood flow to the lungs.
- **Ventricular septal defect.** A ventricular septal defect is a hole (defect) in the wall (septum) that separates the two lower chambers of the heart — the left and right ventricles. The hole allows deoxygenated blood in the right ventricle — blood that has circulated through the body and is returning to the lungs to replenish its oxygen supply — to flow into the left ventricle and mix with oxygenated blood fresh from the lungs.
- Blood from the left ventricle also flows back to the right ventricle in an inefficient manner. This ability for blood to flow through the ventricular septal defect reduces the supply of oxygenated blood to the body and eventually can weaken the heart.
- **Overriding aorta.** Normally the aorta branches off the left ventricle. In tetralogy of Fallot, the aorta is shifted slightly to the right and lies directly above the ventricular septal defect.
- In this position the aorta receives blood from both the right and left ventricles, mixing the oxygen-poor blood from the right ventricle with the oxygen-rich blood from the left ventricle.
- **Right ventricular hypertrophy.** When the heart's pumping action is overworked, it causes the muscular wall of the right ventricle to thicken. Over time this might cause the heart to stiffen, become weak and eventually fail.

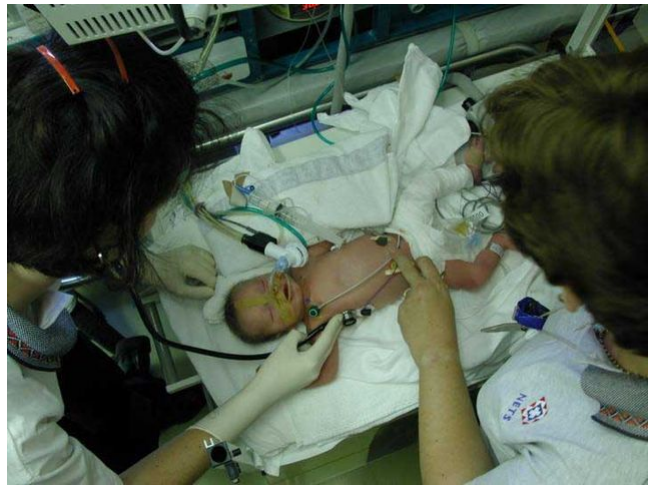
https://www.mayoclinic.org/diseases-conditions/tetralogy-of-fallot/symptoms-causes/syc-20353477

# The 4 Abnormalities:




https://commons.wikimedia.org/w/index.php?curid=860748

# Treatment



- Surgery is the only effective treatment for tetralogy of Fallot.
- Intracardiac repair:
- Usually done within the 1st year after birth, involves several repairs
- Patch over the ventricular septal defect to close the hole between the ventricles
- Repairs or replaces the narrowed pulmonary valve and widens the pulmonary arteries
- The right ventricle will go back to normal thickness (doesn't need to work as hard)
- Temporary surgery:
- May be done if premature birth or pulmonary arteries are undeveloped (hypoplastic)
- A shunt is inserted between a large artery that branches off the aorta and the pulmonary artery
- Intracardiac surgery will be performed when baby is ready and shunt will be removed

https://www.mayoclinic.org/diseases-conditions/tetralogy-of-fallot/diagnosis-treatment/drc-20353482

https://www.pinterest.ca/pin/404690716515378461/

# After Surgery

- **Long-term complications are common:**
- **Chronic pulmonary regurgitation (right ventricle)**
- **Other heart valve problems**
- **Continued leaks after the patch repair, may require a re-repair**
- **Enlarged right ventricle or left ventricle**
- **Arrhythmias**
- **Coronary artery disease**
- **Aortic root dilation (ascending aorta enlarges)**
- **Sudden cardiac death**

https://www.mayoclinic.org/diseases-conditions/tetralogy-of-fallot/diagnosis-treatment/drc-20353482

# Ongoing Care

- **Lifelong care with a cardiologist trained in treating congenital heart disease**
- **Routine follow-up appointments which include physical exam, blood tests, echocardiogram, ECG**
- **Monitor physical activity if there is any pulmonary leakage or obstruction or arrhythmias**
- **Antibiotics for dental procedures to prevent endocarditis**

https://www.mayoclinic.org/diseases-conditions/tetralogy-of-fallot/diagnosis-treatment/drc-20353482

https://www.indiacarez.com/paediatric-cardiac-surgery-treatment-india/

# Coping & Support

- Support groups-provide hope, encouragement and support
- Family physician-provide local resources
- Family & friends-give you a break

- Keep a written record of:
- Diagnosis
- Medications
- Surgeries and dates
- Cardiologist's name and number


https://www.mayoclinic.org/diseases-conditions/tetralogy-of-fallot/diagnosis-treatment/drc-20353482

burrows.nichole28

| Variable | 1980-1985 | 1986-1990 | 1991-1993 | Total |
|---|---|---|---|---|
| Live births | | | | |
| • Total no. | 96,287 | 82,335 | 46,934 | 225,556 |
| • Maternal age <20 yr-% | 13.4 | 11.2 | 10.5 | 12.0 |
| • Maternal age ≥35 yr-% | 4.1 | 6.8 | 8.7 | 6.1 |
| Cases of Down's syndrome | | | | |
| Expected* | | | | |
| • No. of cases | 97.4 | 95.1 | 60.3 | 252.9 |
| • % with maternal age < 35 yr | 80 | 72 | 65 | 73 |
| Ascertained± | | | | |
| • No. of cases | 85.2 | 99.3 | 61.5 | 245.3 |
| • % with maternal age < 35 yr | 79 | 61 | 75 | 71 |
| Cases of Down's syndrome identified | | | | |
| By amniocentesis alone | | | | |
| • No. of cases | 9 | 20 | 10 | 39 |
| • No. of pregnancies terminated | 8 | 19 | 10 | 37 |
| By serum screening | | | | |
| • No. of cases | 0 | 15 | 30 | 45 |
| • No. of pregnancies terminated | 0 | 10 | 27 | 37 |
| • After birth-no. of cases | 78 | 71 | 30 | 179 |
| Reduction in prevalence of Down's syndrome among live births-%¥ | 7.2 | 22.5 | 46.3 | 23.2 |

# STUDENT FEEDBACK

https://docs.google.com/forms/d/1Z0mA72XgmNmnBjfL80KvZqocPp_lD_mu4OSoLVR4VeU/edit

# INSTRUCTOR FEEDBACK

https://docs.google.com/forms/d/1PuFqVc78An82mbj_0NIfMdB3Nh-2_3x5JMNWqWj52Iw/edit