



# Inherited Cardiovascular Conditions

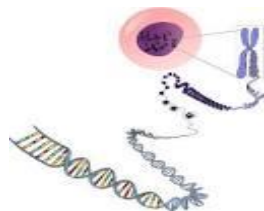
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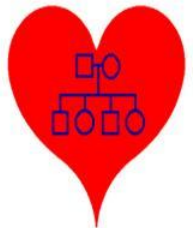
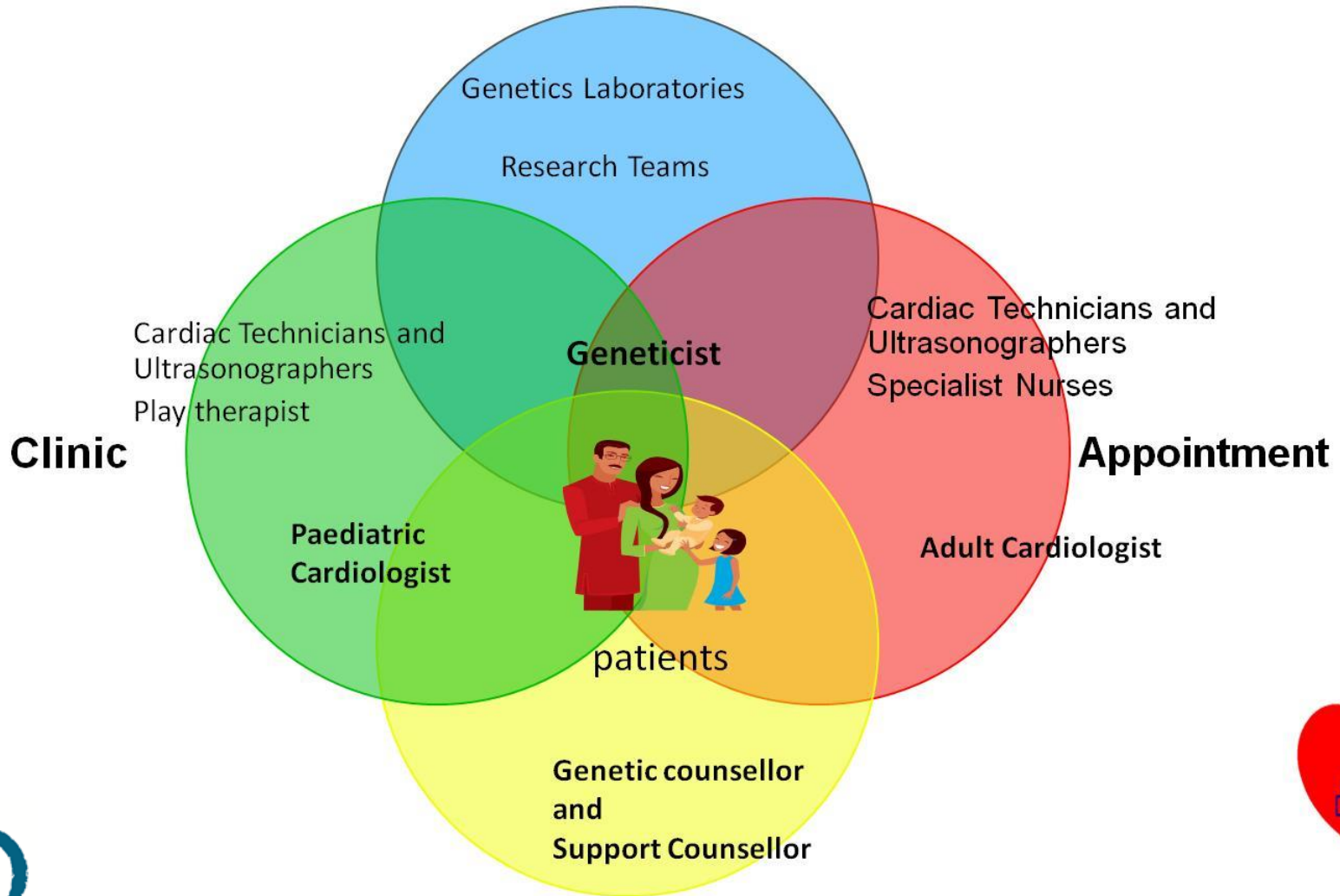


# What are inherited cardiac conditions ?

Inherited or genetic cardiac conditions (ICC), are caused by changes in genes that can be passed from generation to generation. All inherited heart conditions require special care not only for the individual patient but also for their family



# ICC Multidisciplinary Service



# What inherited heart conditions are there ?

## Heart Muscle

### Cardiomyopathies

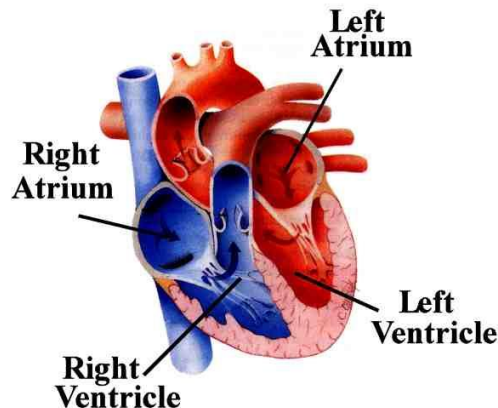
Hypertrophic (HCM)

Dilated ( DCM)

Arrhythmogenic Right Ventricular ( ARVC)

Restrictive

Left Ventricular non –compaction (LVNC)



# What inherited heart conditions are there?

## Arrhythmias (abnormal heart rhythms)

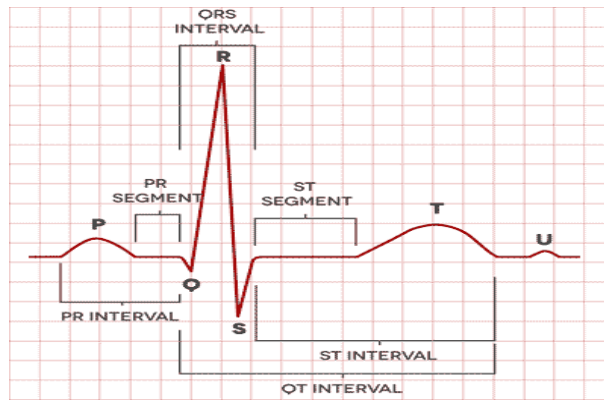
Brugada syndrome

Long QT syndrome (LQTS)

Catecholaminergic polymorphic ventricular tachycardia (CPVT)

Short QT syndrome

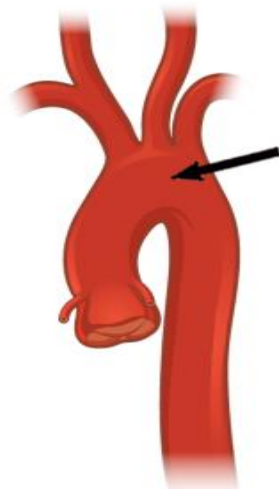
Progressive cardiac conduction defect (PCCD)



# What inherited heart conditions are there?

## Aorta and Vascular

Marfans Syndrome ,  
Dominant Aortic Aneurysms  
Ehlers-Danlos IV ,  
Primary Pulmonary Hypertension  
Loeys-Dietz



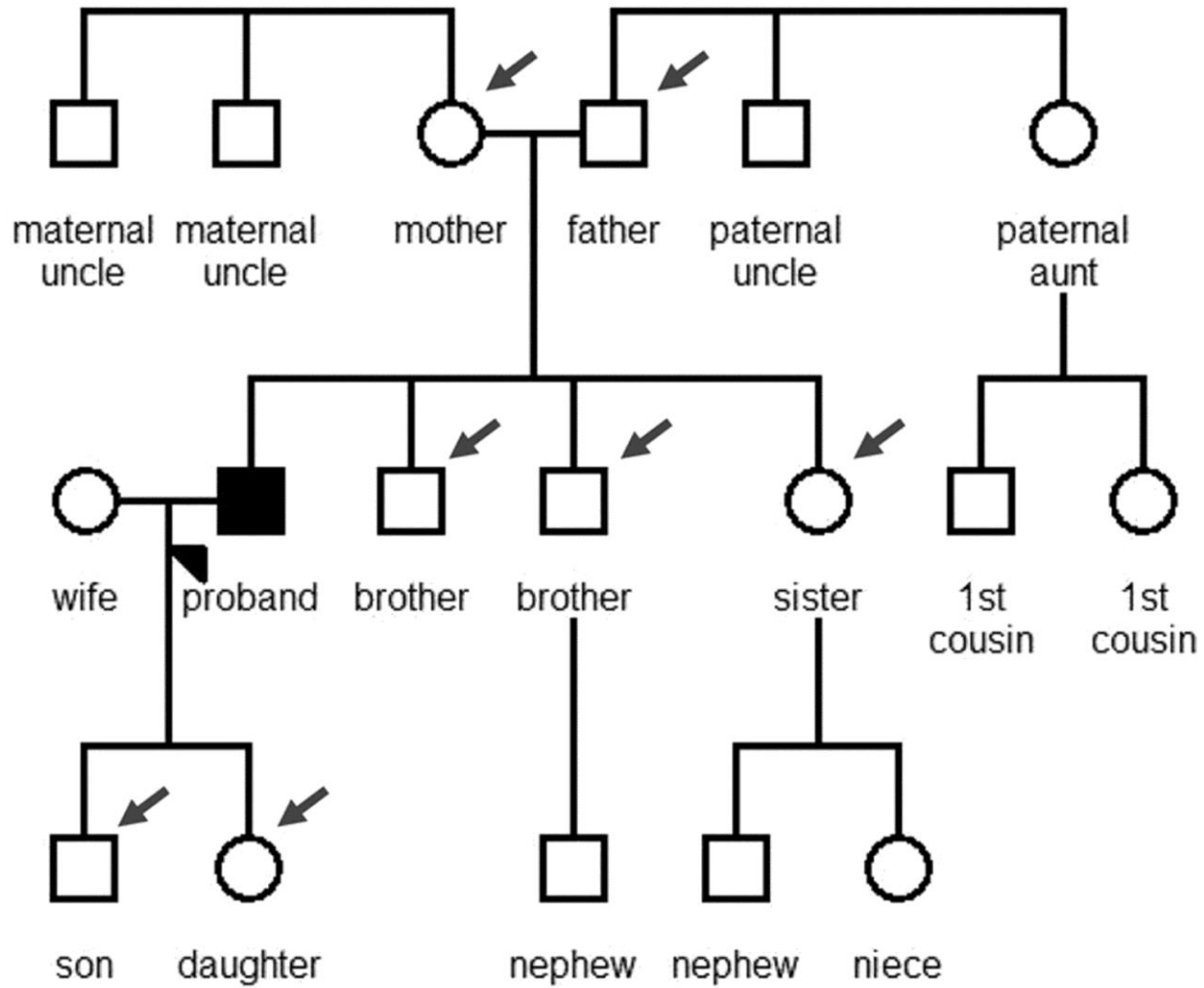
# What inherited heart conditions are there?

Familial Hypercholesterolemia  
Congenital Heart Disorders  
Other genetic conditions i.e. MD

These conditions can have different modes of Inheritance with variable age of onset and severity of symptoms .



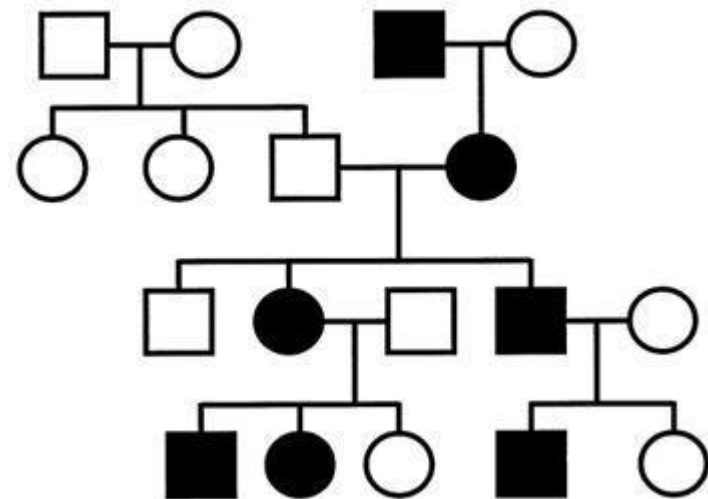
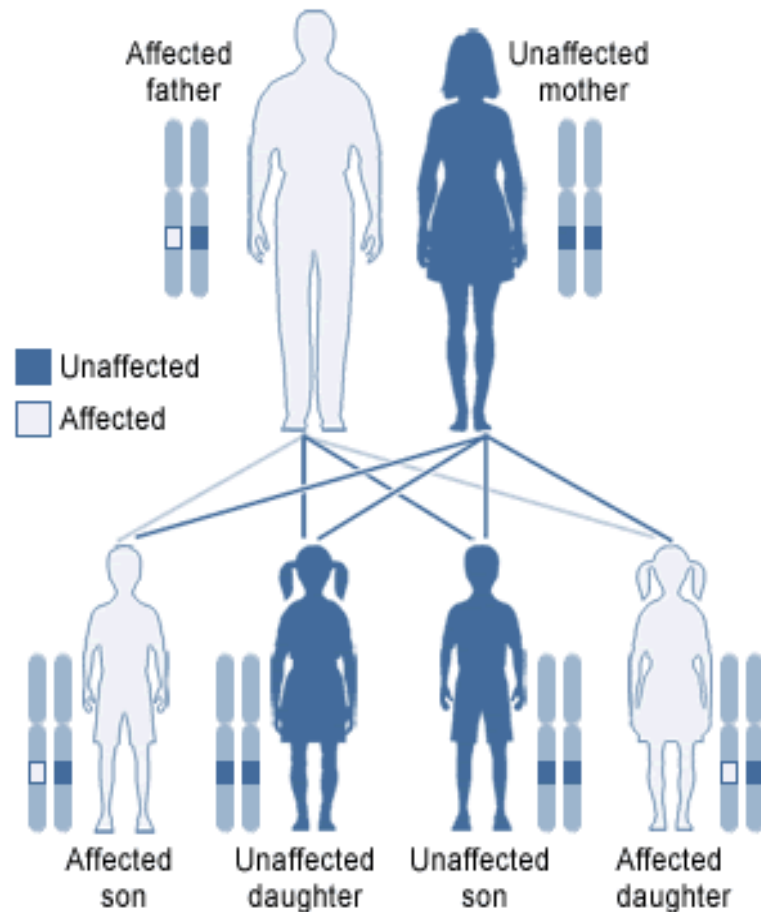
# Pedigree.



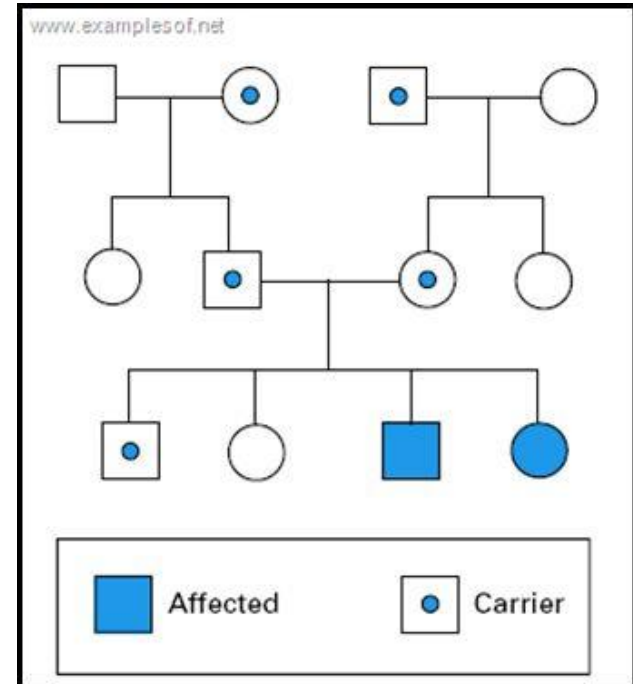
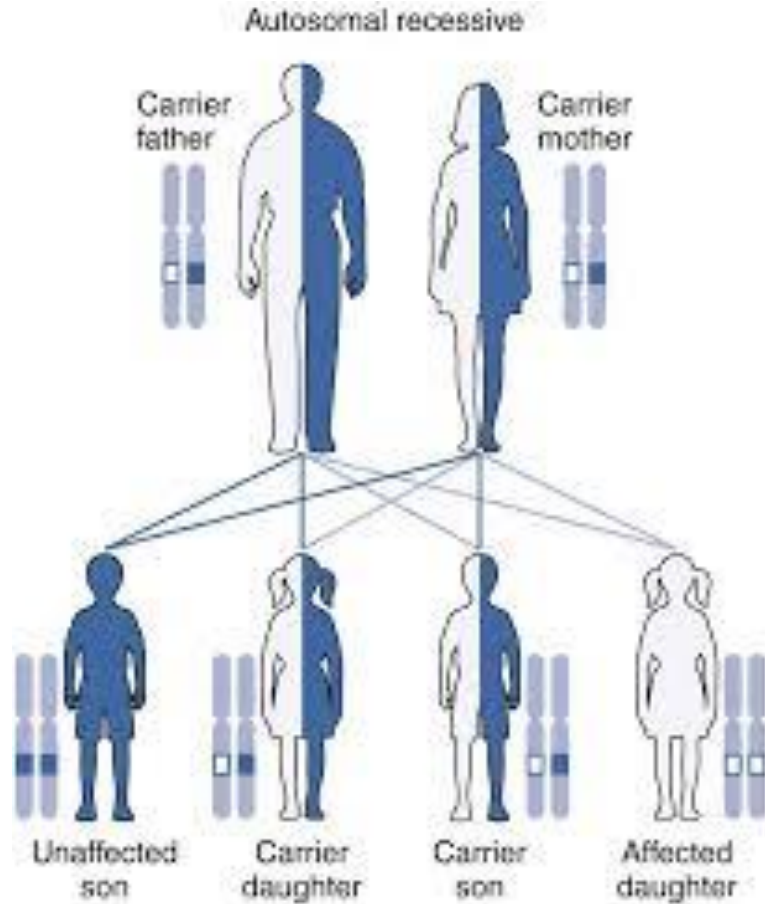


# Dominant Inheritance

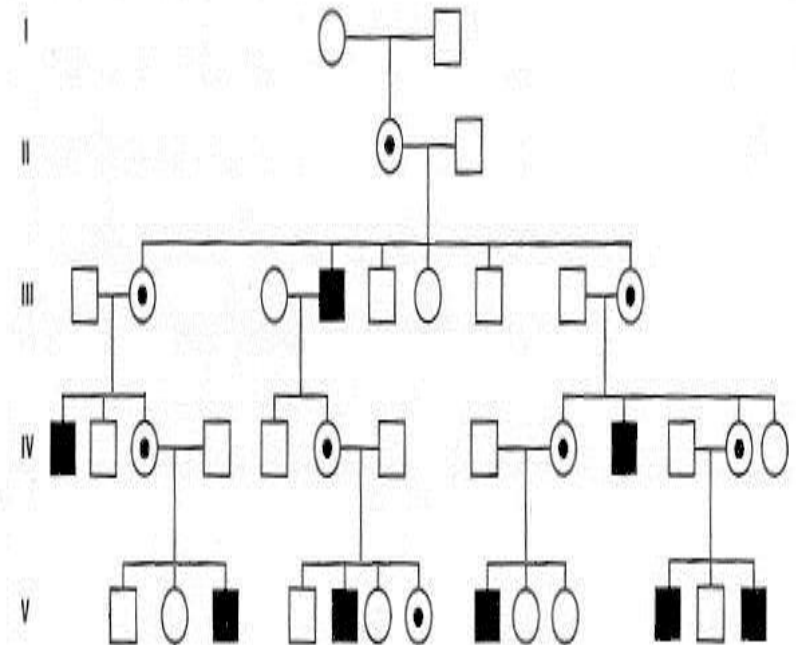
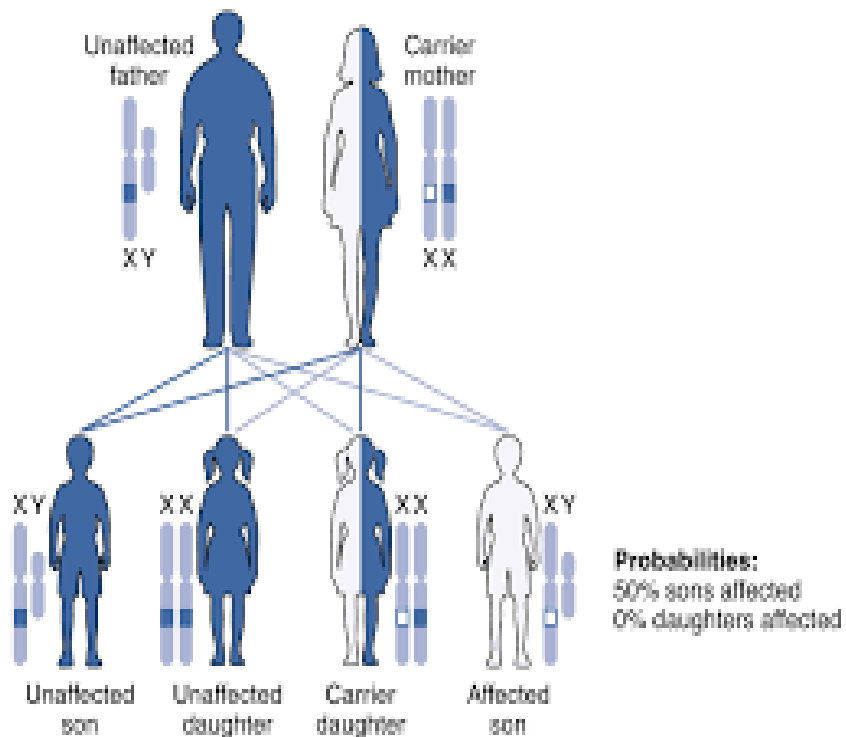
## Autosomal dominant



# Recessive inheritance

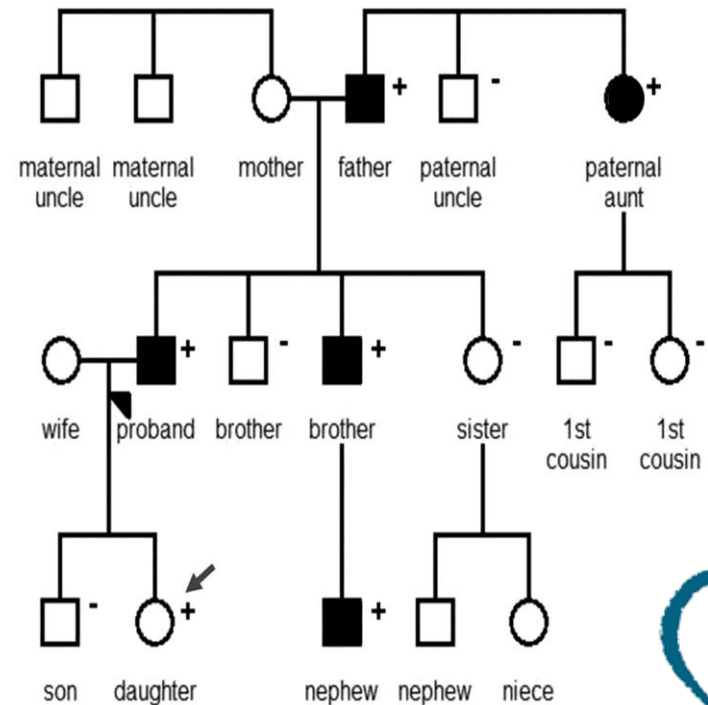


# X-linked inheritance



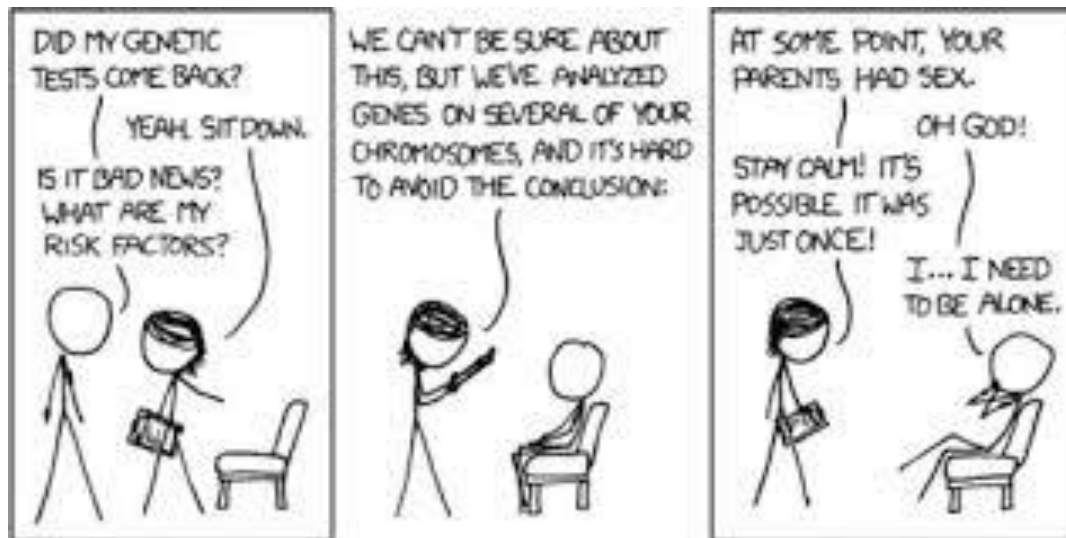
# Screening

All first-degree family members should continue to undergo the screening recommended for the family's heart condition.



# Some basic genetics

- Genes are inherited from our parents
  - half from mum and half from dad
  - We pass half our genes to our children



# Some basic genetics

- Genes are our instruction manuals.
- They tell our body how to grow, develop and function
- They are made of DNA
- Genes code for proteins, which are our building blocks
- We have about 24,000 genes!



# The genetic minefield...

We are usually looking for a 'spelling mistake' in the gene.

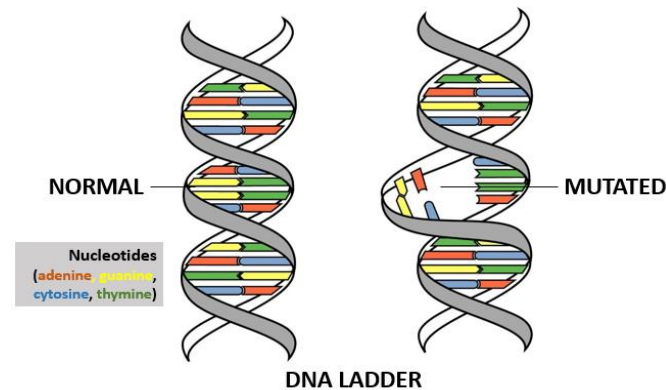
- Genes are read in triplets of letters:
- the big man ran for the bus ...  
the bgm anr anf ort heb us.... (deleted i)  
the big man rag for the bus ...  
the big man x

Important changes in genes are called pathogenic mutations. The term pathogenic means disease-causing. We also get normal variation, and some variants that are difficult to interpret (VUS)



# Gene mutations

Genetic testing usually examines a panel of multiple genes known to cause the specific inherited condition. A single change in 1 gene is sufficient to cause disease. In some cases, 2 or more mutations may be responsible for causing disease in the family.





# Why test?

- Why would a patient want a genetic test?
  - For themselves
  - For their children
  - For relatives



# Diagnostic and predictive testing

- A diagnostic test is done on someone with the condition, to try and locate the altered gene
  - Genetic testing won't change that the diagnosis, but genetic testing is not yet perfect. Even if we don't find the gene responsible, it still may be an inherited condition
  - If we do identify the gene, other (adult) relatives could opt for predictive testing
  - If we don't identify the gene, other relatives can still have screening for but there won't be a genetic test for them yet.



# Diagnostic and predictive testing

Genetic testing can be used:

To clarify the diagnosis in a person who has an inherited heart disease , it may also have some implications for treatment

To offer the option of a predictive gene test to unaffected relatives if they want to know whether or not they are at risk of developing the family's heart condition

To provide options for family planning, including preimplantation genetic diagnosis



# The future, and beyond...

- Gene panels
- Whole exome sequencing
- the 100,000 genome project



# Thank you for your attention

## Any Questions

