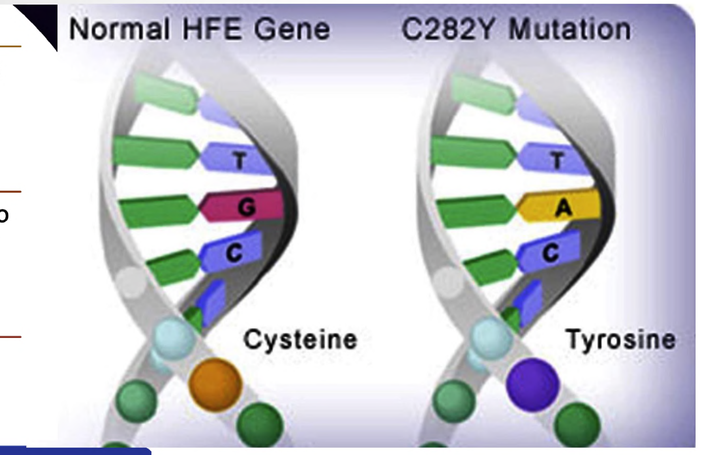
CLINICAL GENETICS IN VARIOUS MEDICAL DISCIPLINES & GENETIC PREDISPOSITIONS AND THE INFLUENCE OF ENVIRONMENTAL FACTORS

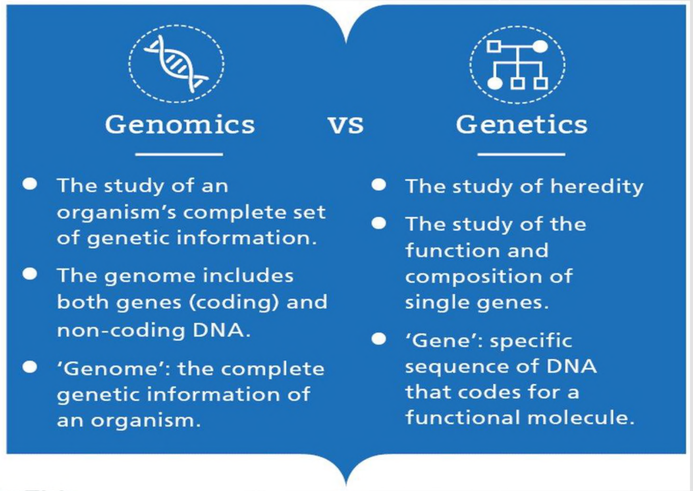
GENETICS FOR DIAGNOSIS BEFORE BIRTH

Advances in techniques allow us to access the diagnosis of some genetic disorders before birth, which help us to treat the disorder or cater to it as soon as possible.

Parental diagnostic screening is one way to allow us to screen a fetus and search for genetic disorders before birth.

Genetic screening can be used to counsel parents about the risks of passing on a genetic disorder to their offspring.





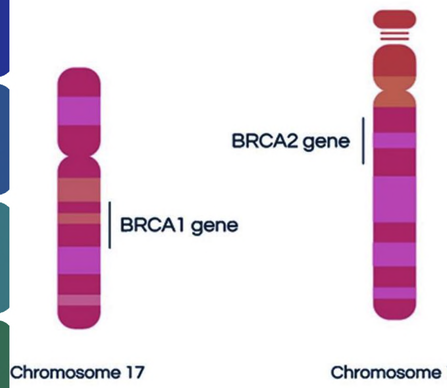
GENETICS AND GENOMICS IN MEDICAL PRACTICE

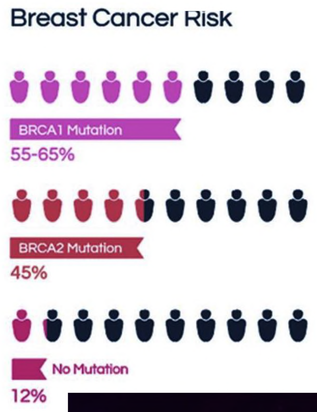
Genetic contribution to medical practice is classified into three categories:

1 – monogenic or chromosomal disorders for example sickle cell anemia, neurofibromatosis or Down syndrome. This is the typical domain of medical genetics and medical geneticists play a huge role in diagnosis and management of such individuals.

2 – more common disorders usually a monogenic cause or single gene mutations cause the disorder. For example breast, ovarian and colon cancer, hemochromatosis and cardiomyopathy. These have incomplete and age dependent penetrance.

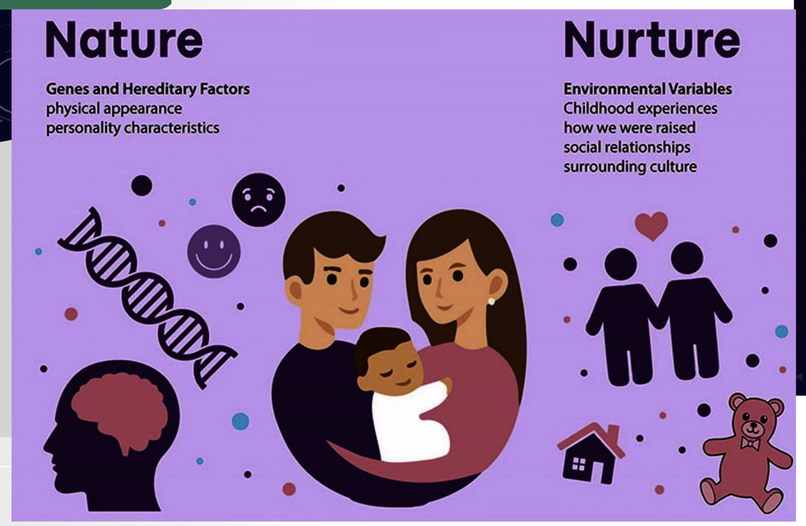
3 – largest and most common category of disorders such as diabetes, hypertension, cancer & cardiovascular disease. Some of these very rarely have single gene causes however mostly it is the genetic contribution





SCHIZOPHRENIA IN GENETICS VS ENVIRONMENT

- Several studies have been provided to prove that environment plays a massive role in activating genes!
- This is seen clearly in various of studies.
- Gottesman found that monozygotic twins have a 48% risk of getting schizophrenia whereas dizygotic twins have a 17% risk rate. This is evidence that the higher the degree of genetic relativeness, the higher the risk of getting schizophrenia.
- However he only proved the importance of genetics.



GENETIC PREDISPOSITIONS AND THE INFLUENCE OF ENVIRONMENTAL FACTORS

Most common diseases are a result of both genetics and environmental factors.

Environmental factors include personal choices such as the food you eat, exercise and external factors such as stress, water quality and air pollutants.

Very little diseases are strictly related to gene mutations but an example of these are Huntington disease and Tay Sachs.