

# STUDY UNIT 1: INTRODUCTION TO GENETICS AND HUMAN HEALTH

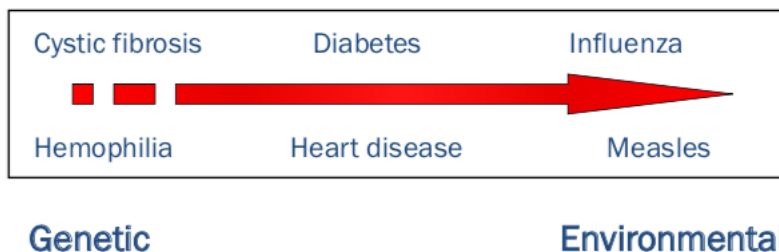
## 1.1) Definitions:

1. **Genetics** : The study of inherited variation
2. **Human Genetics**: Study of inherited human variation
3. **Medical Genetics**: Study of genetic variation related to health and disease

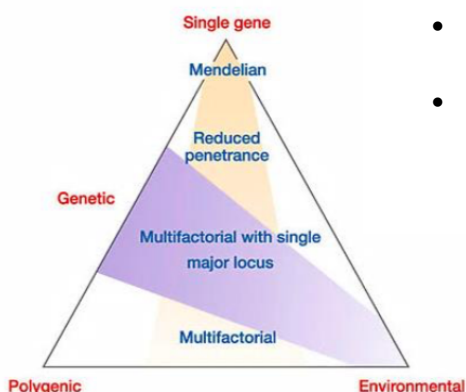
## 1.2) Classification of genetic disorders:

- Chromosomal-Change in the number or structure of chromosomes (Down syndrome)
- Single-gene – When a certain gene is known to cause a disease ( Cystic Fibrosis)
- Multifactorial – Diseases caused by many contributing factors (heart disease and diabetes)
- Acquired somatic disease - Mutations acquired in person's a lifetime (Mutations in an organ or cell – cannot be passed on to offspring)

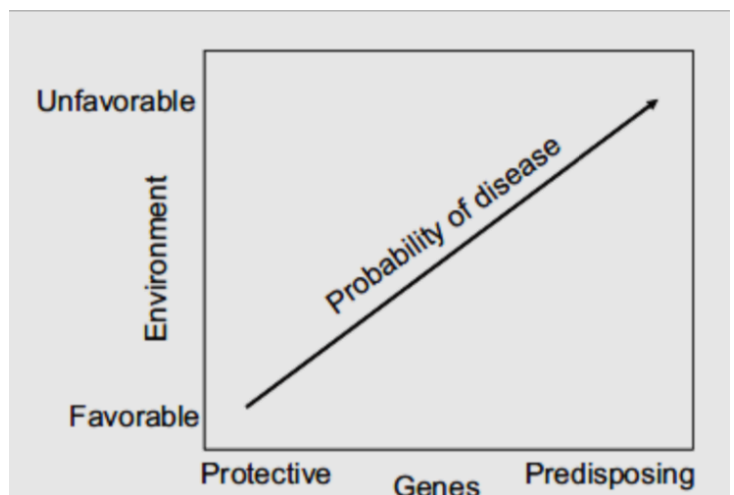
## 1.3) Human disease along a continuum



- Environment = internal (food, smoking, exercise) and external (pollution)
- Background genetics protects you against exposure = Certain genotypes reduce susceptibility to environmental factors (This is often ignored)



- Few characters are purely Mendelian , Polygenic or Environmental
- There is a mix of major and minor genetic determinants + environmental influences
- There are a mix of factors determining any given character represented by a point somewhere in the triangle



Phenotype = Genotype x Environment x Time

#### 1.4) What Is the clinical impact of genetic disease?

- We're basically asking what fraction of people in the population will be found to have a genetic related disorder
- Before and after age 25 : Multifactorial genetic disorders contribute the most to ill health

Category	Number / 1000
Single-gene	3,6
Multifactorial	46,4
Chromosomal	1,8
Undefined	1,2
<b>Total</b>	<b>53</b>

Diagram to left : Genetic contribution to ill health before age 25

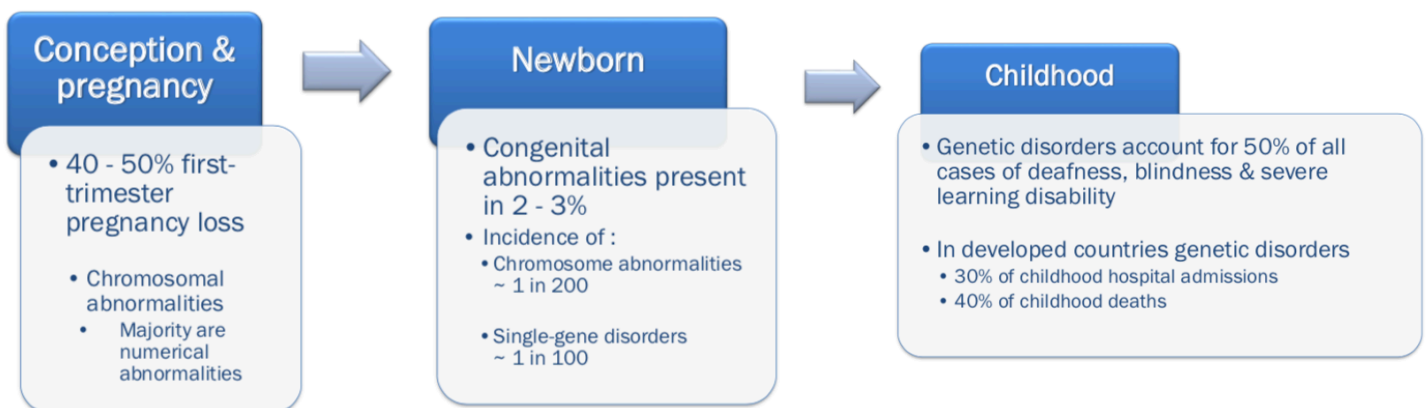
- Roughly 5% of population will have a disorder that can be attributed to "genetic factors"

Category	Number / 1000
Single-gene	16,4
<b>Multifactorial</b>	<b>600</b>
Chromosomal	2
Somatic cell genetic disorders (cumulative)	240
<b>Total</b>	<b>858</b>

Diagram to left : Genetic contribution to ill health after age 25

- Roughly 85% of population will have a disorder that can be attributed to "genetic factors"

#### Morbidity and mortality due to genetic disease:



- First trimester lost – Trisomy lead to spontaneous loss of the pregnancy
- Newborn – Some have clinical presentation at birth, others only appear later (eg facial dysmorphism, sucking reflex)

#### Somatic cell genetic disorders - Normal mutations accumulate

- Most problems in pregnancy are chromosomal abnormalities
- Inborn errors of metabolism = baby cannot thrive and struggle to survive (mutation that occurred during development = genetic)
- Diseases of complex origin arise due to the environment and genes