

Mendelian Disorder

1. Autosomal Dominant	2. Autosomal Recessive	3. X-linked Recessive
- Both sex involved	- Both sex involved	- Males only
- Generations not skipped	- Generations skipped	-

Single Gene Disorder

Enzyme Defects

1. Accumulation of substrate
2. Lack of product
3. Failure to inactivate a protein which cause damage

Structure protein defects

1. Marfan Syndrome
2. Ethler-Danlos Syndromes

Receptor protein defect

- Familial hypercholesterolemia (AD)

Mendelian Disorder

Autosomal Dominant	Autosomal Recessive	X-link recessive
- Both sex involved	- Both sex involved	- Males only
- Generations not skipped	- Generation skipped	- Generation skipping doesn't matter

Chromosomal Disorder

- Chromosome, genome mutation

Down Syndrome

- Meiotic nondisjunction of chromosome 21

Klinefelter Syndrome

- ≥ 2 X chromosome/ $1 \geq$ Y chromosome
- 47,XXY
- Testicular abnormality

Turner syndrome

- Complete/ partial monosomy of X chromosome in females
- Neck webbing, heart defects



By **damn**
cheatography.com/damn/

Not published yet.
 Last updated 9th December, 2023.
 Page 1 of 1.

Sponsored by **ApolloPad.com**
 Everyone has a novel in them. Finish Yours!
<https://apollopad.com>