

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	Structure protein defects
1. Accumulation of substrate	1. Marfan Syndrome
2. Lack of product	2. Ethler-Danlos Syndromes
3. Failure to inactivate a protein which cause damage	
	Receptor protein defect
	- Familial hypercholesterolemia (AD)

Mendelian Disorder		
Autosomal Dominant	Autosomal Recessive	X-link recessive
- Both sex involved	- Both sex involved	- Males only
- Generation 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



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