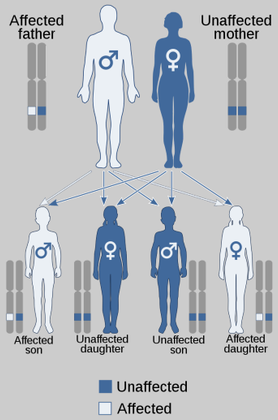
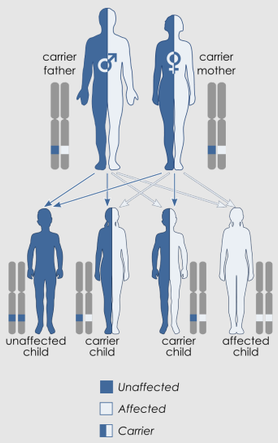
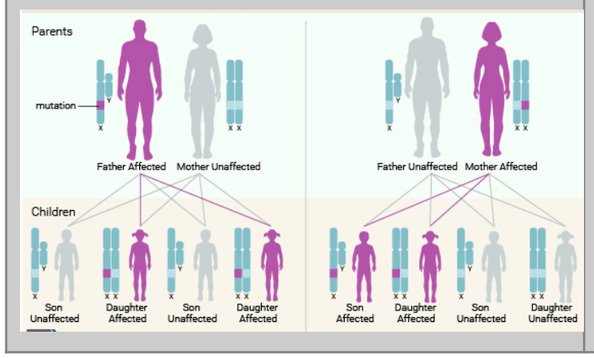
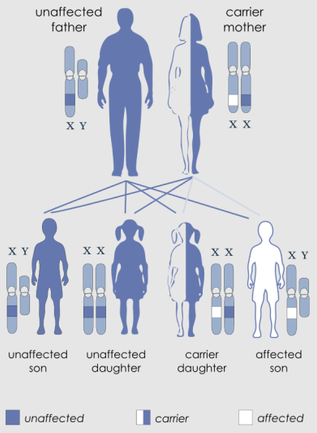
Mendelian Inheritance Patterns	Incidence	Characteristics	Recurrence Risk	Examples
<u>Autosomal dominant</u> 	1/200 individuals	• Wide variation of expression • Vertical transmission of disease phenotype • No skipped generations • ♂ and ♀ equally likely to transmit to offspring	For offspring of one carrier parent, recurrence risk is 50% If both parents are affected, recurrence risk is 75%	Very Powerful **DOMINANT** Humans • **V**on willebrand disease/**V**on hippel-lindau • **Pseudohypoparathyroidism** • **D**ystrophia myotonica • **O**steogenesis imperfecta/ **O**sler-weber-rendu • **M**arfan syndrome • **I**ntermittent porphyria • **N**eurofibromatosis • **A**chondroplasia /**A**dult polycystic kidney disease • **N**oonan syndrome • **T**uberous sclerosis • **H**ypercholesterolemia • **H**untington's disease • **H**ypertrophic obstructive cardiomyopathy • **H**ereditary spherocytosis • **H**ereditary non polyposis coli • **H**ereditary hemorrhagic telangiectasia
<u>Autosomal recessive</u> 	Rare in population	• Less variation in expression than autosomal dominant diseases • Clustering of the disease among siblings • ♂ and ♀ are equally likely to transmit the disease to offspring	For offspring of two carrier parents, recurrence risk is 25% If an affected homozygote mates with a heterozygote, recurrence risk is 50% If both parents are affected homozygotes, recurrence risk is 100%	• Sickle cell anaemia • Cystic fibrosis • Tay-Sachs disease • Phenylketonuria • Mucopolysaccharidoses • Lysosomal acid lipase deficiency • Glycogen storage diseases • Galactosemia
<u>X-linked dominant</u> Rarer than X-linked recessive diseases 		• Twice as common in ♀ than in ♂ • Fathers cannot transmit the disease to sons • Rare to have skipped generations • Heterozygous ♀ may be less severely affected than affected ♂ • An affected ♀ has a 50% chance of passing the disease to her sons or daughters	Dependent on genotype of each parents and the sex of their offspring	• fragile X syndrome

<u>X-linked recessive</u>  	More common than X-linked dominant diseases	• Since ♀ have two copies of the X chromosome and ♂ have only one, X-linked recessive diseases are usually clinically evident in ♂ • Fathers cannot transmit the disease to sons • Can have skipped generations • An affected father will transmit the disease to all of his daughters, who will be carriers and pass the disease onto ~ half of their sons	Dependent on genotype of each parents and the sex of their offspring	• Hemophilia • Fabry disease