Missing topics from the sheets

Degrees of dominance

(depends on which level you are interested in phenotypic level , biochemical level or molecular level and so on ... remember that each gene has TWO alleles (allele is a variation in a gene's nucleotide sequence))

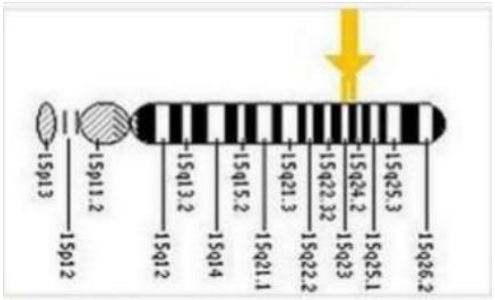
- **Complete dominance (السيادة التامة) :** occur when phenotype of the heterozygous and the dominant homozygous are the same (the presence of one dominant allele is enough to cause its phenotype)
- **Incomplete dominance (السيادة غير التامة):** the phenotype of the daughter is a hybrid in between the phenotypes of the two parental varieties (a red rose and a white rose will produce a pink rose)
- **Co dominance (السيادة المشتركة) :** two dominant alleles both are expressed so affecting the phenotype in separate, distinguishable ways (red rose and pink rose if both are dominant they will produce a rose with red spots as well as white spots)

every trait can be dominant (سائدة) or recessive (متنحية)

tay-sachs disease

One of the lysosomal storage disease.

Cause: mutations on the chromosome 15 in the HEX A gene which produce a lack of Hexoaminidase A (since this mutation causes under expression of this gene).



The HEXA gene is located on the long (q) arm of chromosome 15 between positions 23 and 24.

Consequences: Hexoaminidase A is important for lipid metabolism so mutations will lead to non-functional lysosomes (contain hydrolytic enzymes) and accumulation of lipids in the neurons.

-symptoms usually appear after several months from birth, these symptoms include loss of motor control, blindness, seizures and paralysis and they usually die after a few years from birth.

Features: are in its spelling: T > Testing recommended. A > Autosomal recessive (so one mutant allele isn't enough to cause the disease and the person is a carrier in this case) .Y > Young death (<4yrs). S > Spot in macula (cherry red spots).A > Ashkenazi Jews (usually occur in them). C >CNS degeneration .H > HEX A deficiency. S > storage disease.

What is the degree of dominance in TAY-SACHS:

-**At the organismal level (diseased or carrier or normal)**, the allele is recessive so the normal allele is complete dominant.

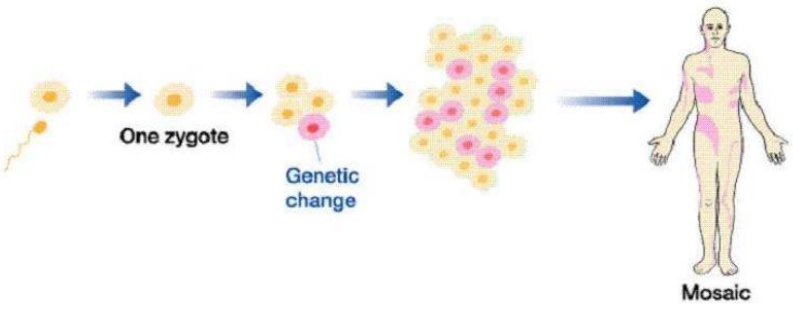
-**At the biochemical level**, (i.e., the enzyme activity level) is incompletely dominant since in heterozygous (a carrier) it will have functionality of the enzyme between the normal and affected.

-**At the molecular level (on the DNA expression level)**, the alleles are co dominant (both are expressed).

if you were asked without determining the level you say it is complete dominance in regard for the normal allele*

Mosaicism and chimera

- **Mosaicism : occurrence of two or more distinct cell lines in the same person that develops from one zygote** due to nondisjunction (during mitosis) or mutations (point mutations , mistakes in DNA replication)or chromosomal abnormalities ... if this occur in one cell this mutated cell will produce more and more population of cells with the same mutation .. So the challenge here is if you take a blood sample for example you won't find a mutation but if you take from the affected tissue you will see it why? because in this example the progenitor blood cells were normal so they produce normal blood cells while the opposite happen in the affected tissue) .. the mosaic form of a disease is less severe than a complete disease (ex. Mosaic down)



- **Chimera: occurrence of more than one populations of cells in the same person that develops from two zygotes** which were fused together since they are closed to each other forming an embryonic issue.

 Remember if those two zygotes develop in different areas in the endomatrium the will result in non-identical twin if they were close to each other there will be a chimera.