



Genetics

Lecture 3:

1) Penetrance → e.g. Waardenburg syndrome

Symptoms → Heterochromia / displacement of the inner canthi / white forelock / deafness

* Only 20% of people with this disease are deaf (Reduced penetrance)

2) Variable Expressivity → e.g. (A) Neurofibromatosis
(B) Myotonic dystrophy

3) Variable age of onset → e.g. Huntington disease

* The onset of the disease is typically in the 40s, however it may range from the 20s - 60s

4) Pleiotropic → e.g. Marfan syndrome

* The syndrome affects the skeletal, cardiovascular & ocular systems

5) Genetic heterogeneity → - Allelic heterogeneity e.g. (A) Duchenne / (B) Becker muscular dystrophy / (C) cystic fibrosis

- Locus heterogeneity e.g. congenital deafness

6) Sex limited → e.g. Autosomal dominant male precocious puberty

7) Sex influenced → e.g. Hemochromatosis

8) Trinucleotide repeats → e.g. (A) Fragile X mental retardation

Symptoms - mental retardation / large head / prominent forehead
& chin / long face / protruding and larger ears / speech delay /
large testes after puberty / loose joints / behavior abnormalities

e.g. hyperactivity / hand clapping & biting / temper tantrums /
sometimes autism spectrum disorder

* most common cause for mental retardation in males

* 30% of males who carry a premutation will develop X-associated tremor/ataxia syndrome (FXTAS)

* 50% of female carriers of a full mutation have mental retardation that is less severe than in affected males

* 20% of females who carry a premature will develop premature ovarian failure (POF)

ⓑ Spinocerebellar ataxia / Ⓒ Huntington disease / Ⓓ Joseph disease
Ⓔ Kennedy disease / Ⓕ Myotonic dystrophy / Ⓖ Friedreich ataxia

9) Imprinting → Ⓐ Prader-Willi syndrome (Deletion is paternally)

* Symptoms → Obesity / voracious appetite / mental retardation

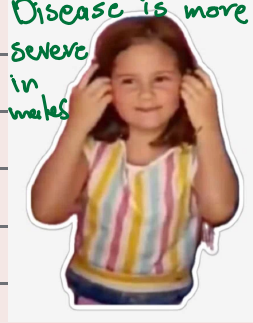
Ⓑ Angelman syndrome (Deletion is maternally)

* Symptoms → Gait ataxia / smiling faces / happy demeanor / mental retardation

10) Inheritance of organelle genes → Ⓐ Mitochondrial myopathy

Ⓑ Leber's hereditary optic neuropathy

Disease is more severe in males



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