

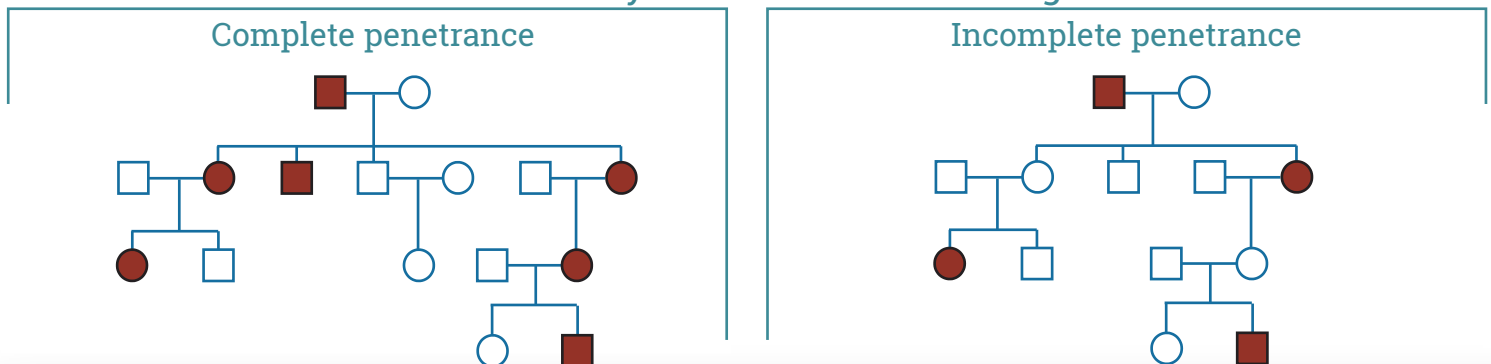


ALS/MND GENETICS

IF I CARRY AN ALS GENE MUTATION, WILL I DEFINITELY DEVELOP ALS/MND?

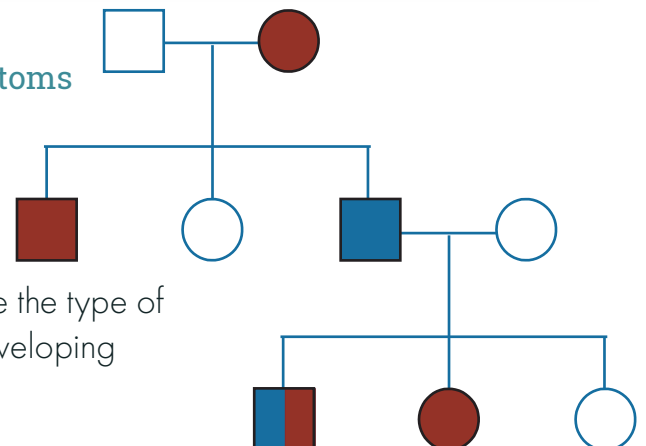
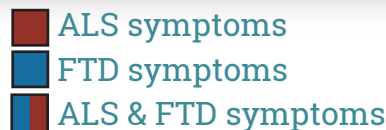
- This is called gene penetrance.
- Penetrance is the risk that you will develop ALS/MND if you carry an associated gene mutation.
- Most ALS/MND gene mutations have incomplete penetrance, meaning there is a chance that symptoms may never develop.
- **Both genetic and other factors (environment, lifestyle, ageing)** contribute to the development of ALS/MND symptoms.

Individuals who all carry the same ALS/MND gene mutation



CAN GENES AFFECT THE TYPE OF ALS/MND?

- ALS/MND varies between individuals:
 - Site of onset of symptoms
 - Age of onset
 - Progression rate
 - Impact on cognition
- Some gene mutations directly cause ALS/MND, and influence the type of symptoms present, while other mutations increase the risk of developing ALS/MND, and modify the symptoms present.



- Some gene mutations are associated with increased risk of developing other diseases as well, such as frontotemporal dementia (FTD) or ataxia. This is called pleiotropy.