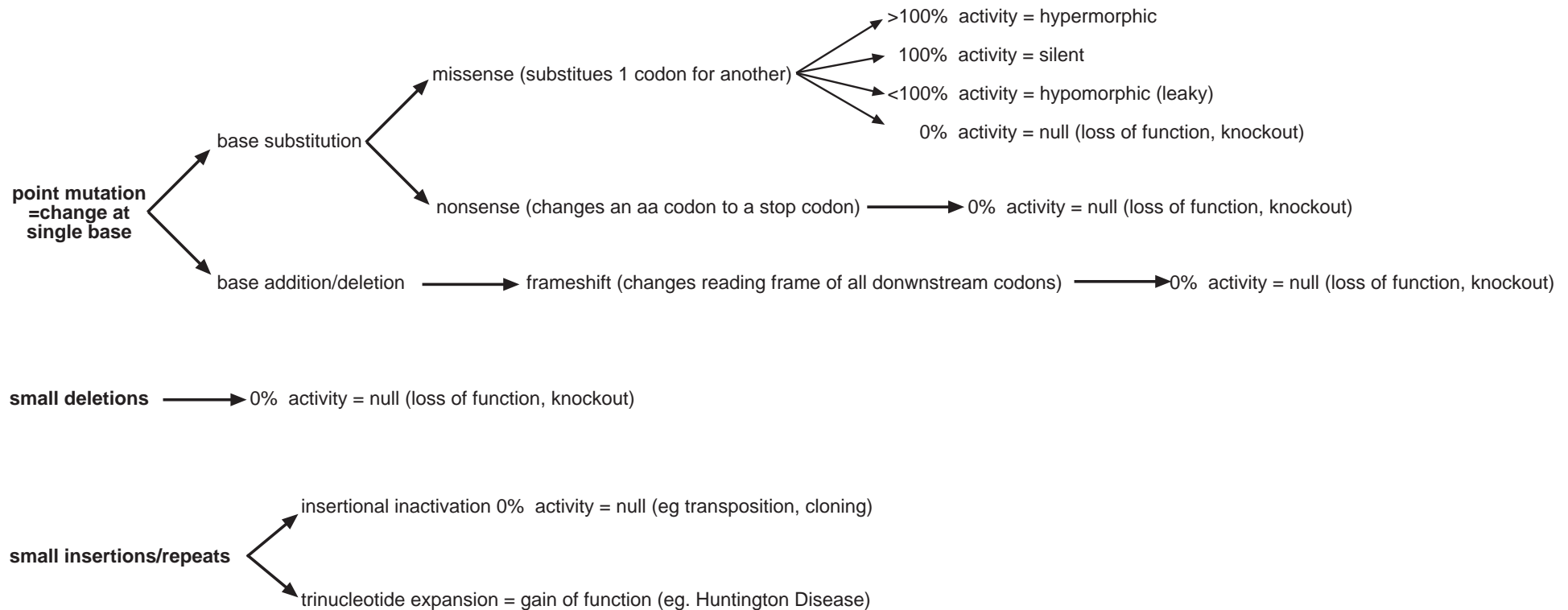


MUTATION = a change within a single gene that creates a new allele



OTHER TYPES OF GENETIC DAMAGE

Multi-gene deletions

- Aneuploidy** {
- trisomy 21 = Down Syndrome
 - trisomy 13 = Patau Syndrome
 - trisomy 18
 - XO = Turner Syndrome
 - XXY = Klinefelter Syndrome

Chromosome Aberrations

- {
- deletions
 - duplications
 - inversions
 - translocations
 - acentric
 - attached chromosomes

ALLELE RELATIONSHIPS

Dominant & Recessive:

A/a = A phenotype = A is dominant and a is recessive:
 Huntington is a dominant mutation: HD/hd = affected
 PKU is a recessive mutation: PH/ph = normal

Semidominance (incomplete dominance):

A/a = phenotype intermediate between A and a
 in snapdragons, A/A = red
 a/a = white
 A/a = pink

Codominance:

A/a = both A and a phenotypes
 A/A = type A blood
 B/B = type B blood
 A/B = type AB blood

loss of function mutations tend to be recessive
 gain of function mutations tend to be dominant