



SIMILARLY FOR DWARVES:

AA } PEAD, Aa } DWARF, aa } NORM, obviously

XD FRAGILE X

FMR1 - expansion of CGG repeats; common in σ
 - chaperons (what??) protect protein folding.

XE HEMOPHILIA

F8C - Def'n's, INVERSIONS
 F9C - Promoter, PT MUT'N
 SYMP: Clotting, excessive BLOOD

XE PMD - DYSTROPHIN IS BL!

- Defect is easy to generate
 Extreme phenos (MOSC waxy)
 Lethal by 18-28
 male BATS

XE Color blindness

etc inability to resist (in gust) - Defect means (receptors)

XE G6PD deficiency

Defect in sugar metab causes Hemolysis
 ICthyosis
 X microdeletion } complex manifestation

Y-LINKED

SRY: 46, XX σ \rightarrow LOC of Y to X - SRY⁺
 \approx Klinefelter

46, XYq \rightarrow LOSS of FUNCTION SRY, SRY⁻

Abnormal gonads \approx tumors affect FERTILITY

NDJ \Rightarrow ANEUPLOID $\xrightarrow{\text{FERTILEN}}$ $2n+1$ (n+n+1)
 MEI I \rightarrow Homol Split
 MEI II \rightarrow SIS Split