Δ in protein seq affects protein fold 2 of 2
Δ in protein fold/structure affects function.
ΔSEQ ⟹ Δ FOLD ⟹ Δ FUNCT

every single cell

This can be bad. Each disease on your list has a *molecular cause*. The following diseases should be marked with a star so you can remember some of the causes and outcomes.

Genetic Disease + (where applicable, Mechanism) (MOLECULAR CAUSES OR DEFECTIVE PROTEINS BOXED)

A DOM ↘

- HUNTINGTONS } Late onset, neurodegeneration, [huntingtin] protein lost
- A. DWARFISM } short stature FIBROBLAST GROWTH FACTOR RECEPTOR 3 [FGFR3] protein FUNCTION LOST.
- POLYDACTYLY / SYNDACTYLY / BRACHYDACTYLY } = many / webbed / stunted } fingers + toes - developmental * we have BRACHY * thumb + toes
- HOLOPROSENCEPHALY } facial + cranial abnormalities, brain damage, [SHH] (Sonic Hedgehog) protein malfunct.

A REC ↘

- ALBINISM } NO pigment in SKIN or hair color - loss of [tyrosinase protein] (see p 164, towle) that forms melanin
- CF } lungs malfunction, dead by 20 w/out lung transplant [CFTR] protein LOST - can't move Cl- (cystic fibrosis transmembrane conductance regulator defect)
- PKU } defect in phenylalanine metab can cause brain damage [PAH] phenylalanine hydroxylase protein malfn
- SICKLE CELL } defect in [HB β-globin] protein assembly - 1 copy confers heterozyg advantage against malaria. HBB encodes β-globin - Val & Glu MUTN causes sickle cell. Thalassemia related.
- TAY SACHS } defect in Nerve demyelination - lethal by age 5 - Hexosaminidase A [HEXA] protein malfunct.

X DOM ↘ ADDED 2013: FRAGILE X } Defect in FMRP - MRNA CHAPERONE - ABSENCE CAUSES CGG DECAY/DAMAGE

- (VITD)-RICKETS } genetically conferred inability to process Vitamin D, Bones soften.
- RETT } severe cognitive delay phenotype / defect in Methyl CpG BINDING PROTEIN 2 [MeCp2] transcriptional regulator.

X REC ↘

- R/G COLORBLIND } condition that knocks out red-green cones IN retina so these colors cannot be plastic guided
- HEMOPHILIA [F8C] clotting factor 8 [F9C] clotting factor 9 / Lack of either of these proteins causes excess. Bleed
- DUCHENNES MD } Defect in [Dystrophin protein] Results in Duchenne or Becker MD. Extreme loss of muscle stabil
- Y LINKED ↘ SRY Defects } [SRY - sex-determining Region Y protein] binds DNA, alters Chromosome Structure. / translocation results in gender defects.