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| PHENYLKETONURIA (PKU) |  | **Change in enzyme that breaks down phenylalanine; Eating foods containing phenylalanine (proteins) causes mental retardation; Low protein diet prevents mental retardation; All babies born in SD are tested for this** |
| SICKLE CELL ANEMIA |  | **Changes in hemoglobin protein causes red blood cells to sickle; causes circulatory problems and organ damage; due to SUBSTITUTION A→T; more common in African Americans; Heterozygous carriers are resistant to malaria** |
| TAY SACHS | C:\Users\riedellke\Desktop\2014-2015\barcodes\Genetic Disorders\TAY_SACHS.png | **Disorder in which lipids accumulate in the brain causing retardation, blindness and early death; More common in Jewish people** |
| ACHONDROPLASIA |  | **Defect in bone formation causing normal sized head/torso, but short arms/legs; Also called “Dwarfism”** |
| HUNTINGTON’S |  | **Brain deteriorates starting about age 30-40;Lose ability to walk, talk, think  Leads to early death; Caused by extra CAG repeats** |
| CYSTIC FIBROSIS |  | **Deletion in ion channel proteins for transporting Cl – ions; Causes thick mucus to clog lungs and digestive organs; more common in Caucasians** |
| DUCHENNE MUSCULAR DYSTROPHY (DMD) |  | **Deletion in gene for muscle protein causing progressive weakening and loss of muscles causing paralysis and eventual death** |
| KLINEFELTER SYNDROME |  | **Males with extra X chromosomes (XXy, XXXy, XXXXy); Infertility; males with some female characteristics; Treated with hormone therapy** |
| HEMOPHILIA |  | **Disorder in which the proteins that clot blood are missing causing excessive bleeding after injuries; found in the royal families of Europe** |
| COLORBLINDNESS |  | **Inability to distinguish the colors  (especially red from green)** |
| TURNER  SYNDROME |  | **X0 females; females have only one X chromosome; infertility; Treated with hormone therapy** |
| DOWN  SYNDROME |  | **Three #21 chromosomes; characteristic facial features; slanted eyes;  mental retardation; some heart defects; Also called Trisomy-21** |