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| GENETIC DISORDER | DUE TO | CHARACTERISTICS |
|  |  | Deletion in enzyme that breaks down amino acid phenylalanine;Build up of phenylalaine causes mental retardationLow protein diet can help prevent retardation |
|  |  | Lack functional enzyme in lysosome to breakdown lipids.Lipids accumulate in brain resulting in seizures, blindness, mental retardation, and early death |
|  |  | Change in code for chloride ion channel protein affects glands that produce mucus, digestive enzymes, sweatAccumulation of thickened mucus in the pancreas, intestinal tract and lungs; Increase risk of bacterial infectionsRelease of excessively salty sweat an indicator of disease More common in Caucasians  |
|  |  | Degenerative brain disease caused by extra CAG repeats on  chromosome #4 ; more repeats = more severe symptomsSymptoms appear in middle age after child bearing years.Irreversible/lethal once deterioration of the nervous system begins; Homozygous dominant = lethal before birth |
|  |  | DD = LETHAL before birth; Heterozygote = shows traitPremature fusion of growth plates in long bones resultsin normal size torso/head and short arms/legs |
|  |  | Substitution mutation (A → T) in hemoglobin geneAbnormal hemoglobin packs together to form rods creating crescent-shaped cells when oxygen is low (crisis) Breakdown of RBC’s- weakness, anemia,Clumping of cells in small blood vessels – pain, organ damage More common in African Americans; also found in Mediterranean/Middle East areasHeterozygote carriers: have s*ickle-cell trait* •produce both normal/abnormal hemoglobin •show malaria resistance (HETEROZYGOTE ADVANTAGE) |
|  |  | Disorder in which the proteins that clot blood are missing causing excessive bleeding after injuries; found in the royal families of Europe |
|  |  | Mutation in gene for protein that detects color; Inability to distinguish red/green most common |
|  |  | Deletion in gene for muscle protein causing progressive weakening and loss of muscles causing paralysis and eventual early death  |

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|  |  | slanted eyes; protruding tonguemild to severe mental retardation; Some have heart abnormalities; Simian crease |
|  |  | *Can be XXY*, *XXXY*, *XXXXY*, *XXXXXY*Males with extra X chromosomesFeminized features/possible breast developmentLack development of male characteristics at pubertyMale sex organs with abnormally small testes; sterile; Usually of normal intelligence; may have learning disabilities;Treated with hormone replacement therapy |
|  |  | Females with only one X chromosomeonly known viable human monosomywebbed neck; broad chest; short statureAt puberty, secondary sexual characteristics fail to develop; internal sex organs do not mature; sterile. |