|  |  |  |
| --- | --- | --- |
| GENETIC DISORDER | DUE TO | CHARACTERISTICS |
|  |  | Deletion in enzyme that breaks down amino acid phenylalanine; Build up of phenylalaine causes mental retardation Low 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, sweat Accumulation of thickened mucus in the pancreas, intestinal tract and lungs; Increase risk of bacterial infections Release 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 symptoms Symptoms 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 trait Premature fusion of growth plates in long bones results in normal size torso/head and short arms/legs |
|  |  | Substitution mutation (A → T) in hemoglobin gene Abnormal 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 areas Heterozygote 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 |

|  |  |  |
| --- | --- | --- |
|  |  | slanted eyes; protruding tongue mild to severe mental retardation; Some have heart abnormalities; Simian crease |
|  |  | *Can be XXY*, *XXXY*, *XXXXY*, *XXXXXY*  Males with extra X chromosomes Feminized features/possible breast development Lack development of male characteristics at puberty  Male 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 chromosome only known viable human monosomy  webbed neck; broad chest; short stature  At puberty, secondary sexual characteristics fail to develop; internal sex organs do not mature; sterile. |