DISEASE	HEREDITY	WHAT WE NEED TO KNOW?
Marphan syndrome	AD	1. Mutation in **fibrillin-1 gene** 2. Affects skeleton, heart and eyes
Enlers-Danlos	AD	1. Deficiency in **collagen synthesis** 2. Hyperplastisity and joint hypermobility
Familial hypercholesterolemia	AD	1. **LDL receptors** dysfunction 2. High cholesterol levels in blood 3. Heart failure and atherosclerosis
Phenylketonuria (PKU)	AR	1. **Phenylalanine hydroxylase** deficiency 2. Phenylalanine accumulation 3. Low tyrosine 4. Mutation in **PAH gene**
Tay-Sacs	AR	1. Ashkenazi jews 2. **Hexosaminidase** deficiency 3. **HEXA** gene mutation 4. High **GM2 gangliosides** 5. Lysosomal defect 6. Cherry red dots (eyes)
Gaucher syndrome	AR	1. Ashkenazi jews 2. **Beta-glucosidase** deficiency 3. Lysosomal defect 4. Accumulation of **glucocerebrosides** in lysosomes of **macrophages** = Gaucher cells
Neimann-pick	AR	1. Ashkenazi jews 2. **Sphingomyelinase** deficiency 3. Cherry red dots (eyes) 4. Lysosomal accumulation of **sphingomyelins and lipids**
Alpha thalassemia	AR	1. Gene deletion 2. Defective **alpha-globulin** (found in all HEME types) 3. Hemolytic anemia
Beta thalassemia	AR	1. No **beta globulin** – found in **HEME type A1** 2. Hemolytic anemia
Congenital adrenal hyperplasia	AR	1. 21 hydroxylase deficiency 2. Low **aldosterone** and **cholesterol** levels
Leucinosis/maple syrup urine disease	AR	1. **BCKA D.H** deficiency 2. Accumulation of **branched chain keto acids** 3. Accumulation of **branched chain amino acids** 4. Caramel smell to urine
Tyrosinemia I	AR	1. **FAH** deficiency 2. High levels of **tyrosine and its derivatives** 3. Liver failure, enlarged kidneys, cabbage smell

Albinism (achromatosis)	AR	1. **Tyrosinase** deficiency 2. Low **melamine** levels -> no pigment 3. Damaging **photoreceptors** -> vision problems
Alkaptonuria (AKU)	AR	1. **HGD** deficiency 2. High **homogentisic acid** 3. High **alkapton** (brown/black) -> dots 4. Black urine
Homocystinuria	AR	1. Low **CBS** 2. High levels of **homocysteine and methionine** 3. Can be detected through urine 4. Affects eyes, skeleton, blood vessels, brain
Ornithine transcarbamylase	**XR**	1. **Ornithine transcarbomylase** deficiency 2. High ammonia levels (affects urea cycle) 3. Vomiting, weakness after protein consumption coma
Galactosemia (classical)	AR	1. GALT deficiency 2. High levels of galactose and derivatives 3. Hyperbilirunemia
MCAD (medium chain acyl-CoA DH)	AR	1. Acyl-CoA DH deficiency 2. Lead to acidosis 3. Hypoglycemia 4. Diet based on carbohydrates and MCFA
Smith-lemli opitz	AR	1. Endogenous cholesterol synthesis deficiency 2. DHCR-7 enzyme 3. Complex disabilities, micropenis
MELAS	Solely maternal	1. Mitochondrial disorder 2. Eyelidptosis, lactic acidosis, stroke 3. Ragged red fibers
LHON	AR	1. Retinal ganglion cells 2. Central vision loss
Hurler syndrome	AR	1. IDUA enzyme deficiency 2. Lysosomal storage disease 3. High mucopolysaccharides = mucopolysaccharidosis I 4. GAG found in urine
Porphyria	AR/AD/XR	1. HEME metabolism 2. Early enzymes deficiency – high ALA, high porphobilinogens 3. Late enzymes deficiency – high porphyrinogens 4. Lead to anemia and photosensitivity – vampirism