PKU phenylketonuria	Maple syrup Urine Disease	Albinism	Homocystinuria	Alkaptonuria
Most common disease of amino acid metabolism.	Deficiency of branched chain α-ketoacid dehydrogenase	Due to **tyrosinase deficiency, Tyrosine and DOPA accumulated.**	Due to defects in homocysteine metabolism	A rare disease of tyrosine degradation
Deficiency of **PAH:** CLASSICAL accumulation of phenylalanine, which results in hyperphenylalanemia &Deficiency of tyrosine	**Classic type:** Most common, due to little or no activity of branched chain α-ketoacid dehydrogenase **Intermediate and intermittent forms:** Higher enzyme activity, symptoms are milder **Thiamine-responsive form:** High doses of thiamine(VitB1) increases α-ketoacid dehydrogenase activity		Deficiency of **cystathionine β-synthase** (Converts homocysteine to cystathionine) leads to homocystinuria / homocysteinemia	Due to deficiency of homogentisic acid oxidase
Deficiency of **BH4:** ATYPICAL Due to deficiency of BH2 reductase, synthetase enzymes			Hyperhomocysteinemia is also associated with: **-Neural tube defect (spina bifida) - (atherosclerosis) -A risk factor of heart disease**	Homogentisic aciduria: -elevated homogentisic acid in urine which is oxidized to dark pigment over time -Arthritis -Black pigmentation of cartilage, --tissue -Usually asymptomatic until adulthood
Conversion of Phe to Tyr requires (BH4) Even if PAH level is normal The enzyme will not function without BH4 → Phe is accumulated				
If BH4 is absent: Tyr will not be converted to catecholamines and Trp will not be converted to serotonin	-The enzyme decarboxylates leucine, isoleucine and valine -These aminoacids accumulate in blood	Tyrosine is involved in melanin production Melanin is absent in albino patients Melanin is a pigment of hair, skin, eyes	High plasma and urine levels of homocysteine and methionine and low levels of cysteine	
Characteristics: -Elevated phenylalanine in tissues, plasma, urine **-Gives urine a mousy odor** -CNS symptoms, -Hypopigmentation.	Symptoms: -mental retardation, physical disability, metabolic acidosis, etc. **-Maple syrup odor of urine**		Symptoms: Skeletal abnormalities, osteoporosis, mental retardation, displacement of eye lens	
Prenatal: by detecting gene mutation in fetus. Neonatal: measuring levels of blood phe. Treatment: Life long phe-restricted diet and tyrosine supplementation.	Treatment: Limited intake of leucine, isoleucine and valine causes no toxic effects	Hair, skin, eyes appear white Vision defects, photophobia	Treatment: Oral administration of vitamins B6, B12 and folate Vitamin B6 is a cofactor of cystathionine β-synthase Methionine-restricted diet	Treatment: Restricted intake of tyrosine and phenylalanine reduces homogentisic acid and dark pigmentation
Enzyme -> Amino Acid Phenylalanine hydroxylase -> Phenylalanine	Enzyme -> Amino Acid α-ketoacid dehydrogenase -> Isoleucine, leucine and valine	Enzyme -> Amino Acid Tyrosinase -> Tyrosine	Enzyme -> Amino Acid Cystathionine β-synthase -> Methionine	Enzyme -> Amino Acid Homogentisic acid oxidase -> Tyrosine and phenylalanine

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