* carbohydrate Disorders

① Galagtosemia
- Autosomal recessive

- defective ⇒ Galctokinase , Galacose epimerase or galctose transferase
- blood biomarkers ⇒ ↓ glocose , ↑ bilirubin , hyperchlorinic metabolic acidosis, normal lactate and pyruvate.

② Glycerol kinase dificiency

- X- linked recessive

- blood biomarker ⇒ Psudo triglyceridemia

- defect in glycerol kinase

③ Glycogen storage diseases.

→ von Gierke (GSD 1A)
- Autosomal recessive
- defect in glucose - 6- phosphate

- blood biomarkers ⇒ Hypoglycemia, lactic acidosis,

↑ urea , libid and triglycerides.

→ pompe disease (GSD 2)

- Autosomal recessive
- defect α 1,4- glucosidase
- blood biomarkers ⇒ NO Hypoglycemia, ↑ CPK

* Amino acids Disorders

1. Phenylketonuria (common in our region)

- Autosomal Recessive
- defected phenylalanine hydroxylase (chromosome 12 q
- blood biomarkers ⇒ elevated Phenylalanine

2. Tyrosinemia (doctor didn't read it's slide so skip)

3. Maple syrup urine disease

- autosomal recessive
- Defect in oxidative decarboxylation of ketoacids
- blood biomarkers ⇒ ↑ leucine, isoleucine and valine
  * presence of Alloisoleucine is diagnostic
- urine organic acids - branched chain 2 keto and 2 Hydroxy acids

4. Glutaric acidemia type 1 (doctor didn't read it's slide)

5. Homocystinuria

- Autosomal Recessive
- cystathionine B synthetase is defected
- blood and urine test for high homocystine and methonine and low cystine.

* Organic acids disorders

① Propionic Acidemia

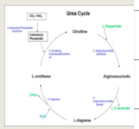
- Autosomal Recessive
- defect in Propyonil CoA carboxylase
- blood biomarkers ⇒ ↑↑ amonia, anion gap metabolic acidosis, neutropenia, thrombocytopenia, hyperglycinemia.

② Isovalitic Acidemia

③ Methylmalomoic Acidemia

* Urea cycle Defects

* just know disease name, enzyme def., mode of inheretance, labs

Urea Cycle Defects	Incidence/ Inheritance	Deficiency	Symptom Onset	Presentation	Labs
CPSDeficiency	1:70-100,000 AR	Carbamoyl phosphate synthetase I	By 5 days of age	Lethargy, hypotonia, vomiting and poor feeding Death if undiagnose d	↑Ammonia ↑CSF Glutamine Respiratory alkalosis Low BUN ↑Glutamine, alanine, asparagine ↓Citrulline ↓Argninine ↓Urine orotic acid 
Ornithine Transcarbamylase (OTC) Deficiency	1:70,000 X-linked (Most common)	Ornithine Transcarbamylase	24-48 hours	Lethargy, hypotonia, vomiting and poor feeding Death if undiagnose d	↑Ammonia ↑CSF Glutamine Respiratory alkalosis Low BUN ↑Glutamine, Alanine, Asparagine ↓Citrulline ↓Argninine ↑Urine orotic acid
Citrillinemia	AR	Arginoinosuccinate synthetase deficiency	Late onset; preceded by stressor		↑Ammonia ↓Argninine
ArgininosuccinicAciduria	AR	Argininocuccinate lyase deficiency	Late onset; preceded by stressor	Trichorrhexis nodosa Episodic coma	↑Ammonia ↓Argninine
Argininemia	AR	Arginase deficiency	Late onset; preceded by stressor	Progressivesplastic diplegia, tremor, ataxia	↑Ammonia Normal Argninine

* Hyperlipoproteinimias :-

Type I Hyperlipoproteinemia	Lipoprotein Lipase Deficiency Increased Chylomicrons and VLDL Hypertriglyceridemia
Type II a Hyperlipoproteinemia	Defect in LDL Receptors Increased LDL levels in blood Hyperbetalipoproteinemia Hypercholesterolemia
Type II b Hyperlipoproteinemia	Increased production of Apo B Increased production of VLDL and impaired LDL catabolism Increased VLDL and LDL
Type III Familial Dysbeta Lipoproteinemias	Defect in ApoE Broad Beta Disease Increased IDL
TypeIV Hyper-pre-β-Lipoproteinemia	Impaired VLDL metabolism. Increased VLDL Due to acquired conditions: ❑ Obesity ❑ Alcoholism ❑ Diabetes mellitus
Type V Combined Hyperlipoproteinemia	Increased VLDL and Chylomicrons Due to acquired conditions: ❑ Obesity ❑ Alcoholism ❑ Diabetes mellitus