Aminoacidopathies

Disorder	Enzyme Blockade	Accumulated Substrate(s)	Presentation	Treatment
Tyrosinemia	Fumaryl-acetoacetase (fumaroaceto-acetate, → fumarate + acetoacetate)	Tyrosine (blood), Succinylacetone (urine)	Liver failure, RTA - due to accumulation of **succinylacetone**	Nitisinone (blocks early step in Tyr metab - can't make succinylacetone), Tyr restriction

Carbohydrate Metabolism

PowerPlans	Galactosemia Admit Orderset
Biochemical Defect	Issues with glucose/fructose/galactose metabolism
Presentation	Timing depends on intro to culprit carb (**galactosemia** early d/t breastmilk, **fructose** introduced later) and from timing of spacing feeds (longer fasting = need to mobilize glycogen stores → GSD becomes manifest); often p/w metabolic crises (lethargy, encephalopathy, HD instability); may have stigmata of toxic deposition (see chart)
Diagnosis	Galactosemia is on the NBS (hereditary fructosuria and GSD are not); definitive with enzyme assays from blood (also done on cultured fibroblasts & liver); suggestive labs = hypoglycemia, ketosis, metabolic acidosis, liver dysfunction; reducing substances in urine present in galactosemia + hereditary fructose tolerance

Disorder	Enzyme Blockade	Accumulated Substrate(s)	Presentation	Treatment
Classic Galactosemia	Galactose-1-phosphate uridyl transferase (allows for transfer of Gal-1-P to Glu-1-P)	Gal-1-P, total galactose + urine **reducing substances**	Hepatomegaly, jaundice, vomiting, cataracts, FTT, lethargy, proximal RTA (Fanconi syndrome), *E Coli* sepsis after starting galactose-containing feeds (e.g., breastmilk).	No **galactose** - includes no **lactose** (milk / dairy)
Hereditary Fructose Intolerance	Aldolase B (splits F-1-P into DHAP + glyceraldehyde)	F-1-P - urine **reducing substances**	Similar to classic galactosemia, but **no cataracts**; occurs w/ fructose-containing foods	No **fructose** from diet - includes no **sucrose** or **sorbitol**
Glycogen Storage Disease (GSD) Type Ia (von Gierke)	Glucose 6 phosphatase (G6P → glucose + Pi)	G6P → lactate, triglycerides, and uric acid	~3-6 months: hypoglycemia 3-4 hrs after meal,, lactic acidosis, hepatomegaly, hypertriglyceridemia, hyperuricemia, "doll face," small size	Frequent meals, Uncooked cornstarch 1.5-2.5 g/kg PO q4-6h, avoid sucrose/fructose/ galactose, $NaHCO_3$ for acidosis, allopurinol for hyperuricemia

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