Inborn Errors in Metabolism

General Labs: Glucose, Ammonia, Lactate, UA
General Management: NPO, MIVF with D10

High Ammonia

Not Acidotic

Urea Cycle Defect

- ↑ Ammonia
- ↓ BUN
- ↑ pH (resp alkalosis)

*OTC deficiency is most common type
*Occurs when you give protein
*Treat with:
  - L-arginine (Catalyzes urea cycle)
  - Sodium Benzoate (nitrogen scavenger)

Acidotic

Organic Aciduria

- ↓ pH
- ↑ Ammonia
- ↓ Bone marrow suppression

*Gluteric Aciduria Type 1: Chronic subdurals, NAT look alike
* Dx: Urine and serum organic acids

Low Glucose

Not Acidotic

Fatty Acid Oxidation d/o

- ↓ Glucose
- ↓ Ketones

*MCAD most common inborn error of metabolism
*Occurs when fasting
* Dx: Serum acylcarnitine profile

Acidotic

Glycogen Storage Dz

- ↓ Glucose
- ↓ pH
- ↑ Lactate
- ↑ Liver size

* Can develop HCC

Galactosemia

- ↓ Glucose
- ↑ Urine Reducing Substances
- ↑ Bilirubin

*Occurs when you give lactose (no more boob)
*E.Coli Sepsis
*Give Abx if presenting sick
*Cataracts develop

Ketotic Hypoglycemia

- ↓ Glucose
- ↓ pH
- ↑ Ketonuria

*Most common cause of low Glucose
*Occurs in toddler to school age kids
*Occurs when fasting

Back to Table of Contents

Crashing Neonate

Hypoglycemia

Testing
Accucheck

Treatment
Give 30 mL of D10W

Response time:
Minutes

Sepsis

Testing
CBC, CMP, BCx, UA, UCx, LP

Treatment
Antimicrobials

Response time:
Hours to days

NAT

Testing
CT head, Bone Survey

Treatment
NeuroSx consult, SW, Forensics consult, Police

Response time:
Immediate to hours/days

Testing
CXR, hyperoxia

Treatment
PGE1
(0.05 mcg/kg/min)

Response time:
Minutes

IEM

Testing
Accucheck, ammonia, pH, UA, CMP

Treatment
NPO, D10W

Response time:
?

Abdominal Catastrophe

Testing
KUB, upper GI

Treatment
Surgery consult

Response time:
Fast

Back to Table of Contents