"Introduction"

(high ammonia) >150 is risky for
Cerebral edema & herniation

Free flowing sample
placed in ice
immediately to the lab.

hypoketotic hypoglycemia < fatty acid oxidation
hyperinsulinism

plasma amino acid used to diagnose

- PKU
- tyrosinemia
- maple syrup
- urea cycle defect
- organic acidemia

Please look in the seminar for (1st par

- How to suspect ?
- clinical scenarios that raise susp
- emergency management.

THE SPECIALTY HOSPITAL المستشفى التخصصي

protein

organic acidemia

① 15 month old, NF, hx of acute weakness lethargy, dec. responsiveness

RR = 45, P 145, 100/59

Na: 140, K 4, Co2 5, Glu: 45, Cl 98
NH3 57.

BG: 7.02, 19, 4.8, urine +ve for ketones

??

* previously healty, with acute onset.
↑ AG, Metabolic acidosis, NL or slight ↑ NH3
(organic acidemia).

- methylmalonic
- propionic
- glutaric acidem

Dx urine & Serum amino acid.
plasma acylcarnitine

Catabolic crisis upon febrile illness with dystonia & subdural hemat

Rx: Start glucose
IV. lipid & no proteins

amino acidopathy :-

maple syrup
- poor arousal & feeding
  loss of moro, seizure
- ↑ leucine
  alloisoleucine.
- ∓ Met. acidosis
  ketosis
- Diet control.

PKU
- most common.
- phenylanine ollase def.
- permenant damage
- fair hair & skin
  eczema
  light sensetivity.
  musty or mousy odor
  epilepsy
- Brain MRI, demylination
- diet restriction
  BH4 supplement.
- monitor phenylalanine level.
- start ttx before 2 weeks.
- if NOT controlled in pregnant female cause syndrome.

Homocystinuria (AR)
- cystathionine-b synthetase

ttx Betaine
methionine restrict
ASA.
B12, folic acid, B6

- ↓ IQ
  ↓ lens.
  Thromboembolism.
  Limited mobility.

Urea cycle defect OTC is xlinked / Cirtullinemia / arginos

⇒ excrete ammonia into urea → out body
always ↑ NH3, Cerebral edema, SZ, low BUN
WO acidosis

ttx: Dextrose, Na-benzoate, dialysis
low protein diet

lipid

Fatty acid oxidation

NB ketotic hypoglycemia, prolonged fasting

~~NAI~~ MCAD → lethargy / SIDS / Reye's syndrome

LCAD → Cardiomyopathy / Rhabdomyolysis

↳ avoid fasting, carnitine supplement

carbohydrate

Newborn with hypoglycemia, prolonged jaundice, coagulopathy, E.coli sepsis & Cataracts
Galactosemia

Dx GALT enzyme activity
glactose 1-p. in blood.
Reducing Sub.

DDx glakokinase def.

Peculiar = learning difficulty. DD. (dev. delay)
Tremor POF (ovarian failure)

- Glycogen storge:

in newborn

Von gierk ds: * hypoglycemia, lactic acidosis, ↑uric acid
GSD (I) neutropenia, hepatomegaly. but NO
liver enzyme elevation, hyperlipidemia

* dull face & puffy cheecks, thin extremities

(the liver is unable to release glucose from glycogen
so it will accumelate ⇒ (hepatomegly)

lack of gluconegenesis ⇒ lactic acidosis & tachypnea.

(hypoglycemia is NOT responsive to glucagon)

they need cont. glucose input (corn starch, or NG tube)

Mcardle dx: . muscle cramps post sternous exercise

. ↑ CPK, Myoglobinuria

. avoid sternous exercise

Pompe dx: hypertrophic cardiomyopathy, muscle weakness, ↑CK

- Fructose intolerance: : AR

. 6 months

. accumelation of fructose leads to:

hepatomegaly, vomiting, lethargy, ↓Σ, irritability.

. avoid fructose, sucrose & sorbitol.

Lysosomal storage dz. (progressive dz).
- lack of certain enzymes that break down GAGs in lysosome

- large molecules will be accumelated in vulnerable tissues (mucopolysaccarides, sphingolipids)

↓↓

Hepatomegaly, Splenomegaly, dev. regression
Bone deformities

ex. ① (Hurler, Hunter, Sanfilippo synd.
AR, corneal clouding
x-linked

② Guacher dz — Fabry's dz.

β-glucocerbrosidase def.
- Anemia
- Thrombocytopenia
- Splenomegaly.

Peroxisomal disorder < Adrenoleukodystrophy.
Zellweger synd.

(look for slides)!

Mitocondrial dz: M E L A S
mitoch. encephalopathy — Lactic acidosis — Stroke-like episod

theses Notes are considered as high yeild & guide for reading the seminar itself.

Best of Luck

Dr. Randa Al-Qaisi