

What makes it Metabolic?

- Inborn Errors of Metabolism are individually rare but collectively numerous.
- Latest estimates are 1: 1000 individuals with some IEM.

This depends on age of the individual and the ethnic group.

We have no idea what disorders will be found in South Africa, so pilot studies are needed.

Metabolic Talk

- Clues from the history:
- A previous sibling may have presented in the same manner.
- The parents may be related. AR.
- Similar episodes may have occurred before, yet the child is well between them.
- Response to standard treatment eg of acidosis, hypoglycemia, is poor.

Metabolic Talk

The urine may have an unusual smell, not just ammonia, but sweet or sweaty feet etc.

Other organ systems may be affected-the heart, liver, brain in an unexpected manner, suggesting multi organ involvement.

Presentation can be at any age, but most often and most severe in the newborn.

As many as 25-30% die as newborns in their first episode, called sepsis or brain damage.

Metabolic Talk

- Presentations can be:
- Acute, severe in the newborn: hypoglycemia, hypotonia, seizures.
- Metabolic acidosis, ketosis, jaundice, raised ammonia, increased lactate (especially in CSF).
- Cardiac failure and arrhythmias.
- Always take extra serum and urine for later study, and skin biopsies if terminal and undiagnosed.

Metabolic Talk

- Older Child:
- Seizures, developmental delay, dysmorphic features.
- Behavior and learning problems.
- Ataxia, movement disorders.
- Failure to Thrive.
- Unexplained organomegaly.

Useful Approach

- Saudubray suggests thinking:
- Is the condition due to an excess of a toxin?
- (A normal metabolite accumulates and causes damage—PKU).
- Is it due to problems with energy metabolism—glucose metabolism or a mitochondrial disorder.

Metabolic Talk

- Is it due to an abnormality involving a complex molecule?—the lysosomal and peroxisomal disorders, which we will not discuss in this symposium, as our time is limited.
- The following presentations will help us to elucidate these problems and plan treatment and management .