

Resident tips for working with patients with inborn errors of metabolism

Inborn errors of metabolism are not as scary as you think! For most of the disorders, a little common sense is all that is required. Details about many of the IEM are available on common resources like Up to Date so a little quick reading might help you if you are unsure. Also, don't hesitate to contact your attending physician – it is much better to ask a question even in the middle of the night than to make the wrong decision!

Some basic principles that govern IEM:

1. Most IEM result from enzyme deficiencies that cause the patient to be unable to metabolize a particular substrate – protein, glucose, specific amino acids, fat etc
2. The condition will deteriorate if the patient gets too much of the substrate (for example, a protein load from a GI bleed) or too little of a substrate (for example, if they are fasting and then they become catabolic).
3. Suppressing catabolism is a key part of the management of IEM. Practically speaking, this means providing the patient with adequate calories and giving those calories continuously rather than in meal time boluses. For this reason, then, iv lines and NG tubes providing these calories must be considered important parts of patient care – just as important for example, as the iv line providing insulin infusion in a patient with DKA or the iv line providing thrombolytics to a patient with an acute stroke. Similarly, if the patient is getting calories by the GI tract and then vomits, you have to consider the patient as being potentially catabolic as they have vomited up calories that they need and make up these calories some other way. A table to help you calculate how to provide calories is provided below.
4. When patients with IEM deteriorate, the substrates that accumulate (like NH₃) can impair level of consciousness so monitoring of neurological status is required. For this reason, sedative medications should be avoided if possible.

Common calls that you may receive for patients with IEM and suggested actions:

Call	Resident Action
Iv line has fallen out	Replace iv line stat; make up calories lost during time that IV was out over the next 2-4 hours by increasing iv infusion rates
NG/feeding tube has fallen out	Replace feeding tube stat (even in the middle of the night!); make up calories lost during time that IV was out over the next 2-4 hours by increasing iv infusion rates
Enteral feeds that have been ordered are not available	Contact dietician on call for the hospital; dieticians are available even weekends and holidays; while waiting for the feed to come up, make up the calories with protein free oral intake (if patient is not vomiting – eg. Pop, juice, popsicles) and iv solutions (see table below); some hospitals have pre-mixed supplies of a protein free formula called Duocal (1 kcal/ml) – this premixed supply may be obtained at RCH after hours on the special drug shelf in the RCH pharmacy; after hours at VGH, a frozen supply is available in the pharmacy freezer
Need for HS sedation	Avoid this if possible (ie. If the patient is not at risk of harming themselves); sedation simply to have the patient sleep through the night would be inappropriate if you are monitoring the patient's level of consciousness

How can I calculate calories of the enteral and oral solutions being given?

Solution	Kilocalories provided	Kcal infused per hour at given infusion rate	Maximum infusion rate
20% lipids	1000 kcal/500 ml bottle	If lipids being infused at 21 ml/hr, then 1 hour of infusion provides 42 kcal	Try to avoid infusing more than 500 ml in 24 hrs due to risk of pancreatitis
D10W	340 kcal per liter	If D10W being infused at 42 ml/hr, then 1 hour of infusion provides 14 kcal	Infusion rates above 150-200 ml/hr will exceed renal threshold for glucose reabsorption and the patient will just waste the glucose in their urine
Enteral solutions	Most enteral solutions are mixed at 1 kcal/ml	If enteral solution being infused at 50 ml/hr, then 1 hour of infusion provides 50 kcal	Depends on GI tolerance but don't forget that a normal person can easily drink 330 ml can of soda at a sitting so don't be afraid to increase the enteral feed rate temporarily if the patient is not vomiting

Case example:

You are called by the nurse who is looking after a patient with a urea cycle defect. The patient is getting iv 20% lipids at 11 ml/hr, iv D10W at 42 ml/hr and an enteral feed at 80 ml/hr using an NG tube. The patient has vomited approximately 100 ml of the feed. What should you do?

Step 1 – Hold the feed for an hour and give some antiemetics and then try restarting the feed

Step 2 – calculate the calories lost:

- a. 100 ml of enteral feed in vomit = 100 kcal
- b. 80 ml/hr of enteral feed being held for one hour while antiemetics are given = 80 kcal
- c. Total calorie deficit: 180 kcal to be made up over 2-4 hours
- d. Actions:
 - a. Increase lipids from 11 ml/hr to 24 ml/hr for 4 hours – this will provide an additional 104kcal (ie. $24-11=13$ ml of additional lipid $\times 2$ kcal/ml $\times 4$ hours = 104 extra kcal)
 - b. Increase D10W to 100 ml/hr for 4 hours – this will provide an additional ~ 80 kcal ($100-42=58$ ml of additional D10W $\times 0.34$ kcal/ml $\times 4$ hours = 79 kcal)
- e. Resume previous infusion rates once the calorie deficit has been made up