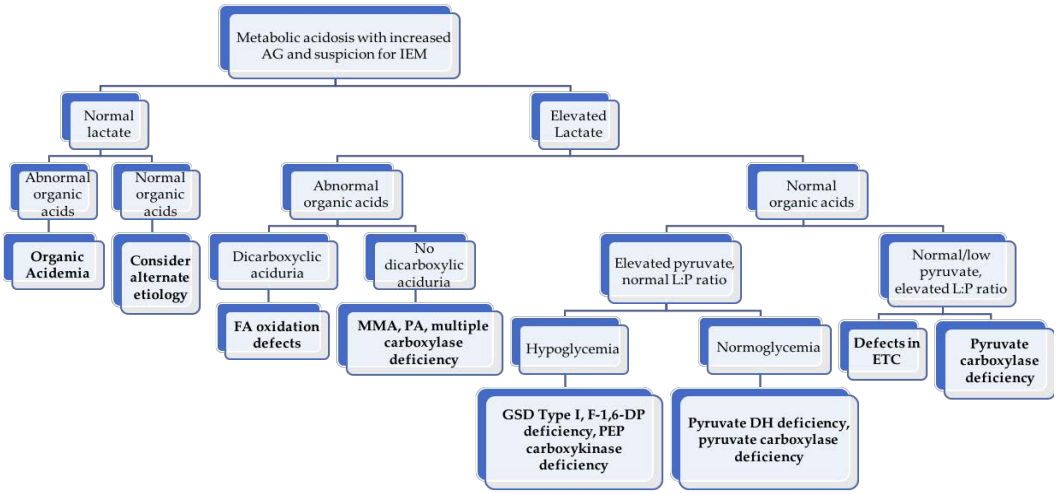
Management of Metabolic Crises

Metabolic Acidosis (when due to IEM)

PowerPlan	Metabolism Lactic or Metabolic Acidosis NOS Admit Plan
Definition	Arterial blood gas with pH < 7.35, pCO2 < 35, bicarbonate < 22
Etiopathogenesis	Inherited: organic acidurias, primary lactic acidemias, renal tubular acidosis; **ANY** metabolic crisis, if left untreated long enough, will progress to metabolic acidosis
Presentation	Acute vomiting, dehydration, lethargy, and rapid, shallow breathing, often h/o protein load
Physical Exam	**Organic acidurias**: limb hypertonia/axial hypotonia, large amplitude tremor, myoclonic jerks, pedaling, sustained paraspinal contraction (opisthotonic posturing) **RTA**: Failure to thrive, polyuria, and rachitic changes **PDH deficiency**: blindness, hypotonia, DD, narrow forehead, frontal bossing, wide nasal bridge, long philtrum, and anteverted nostrils
Treatment	Hydration, caloric intake of 120-140kcal/kg/day, stop proteins initially (esp stop all BCAAs if MSUD is suspected), maintain glucose 100-150 (using high GIR +/- insulin), avoid hypoNa, cerebral edema If serum bicarb < 14 meq/L and pH < 7.2, give IV bolus NaHCO3 as 2.5 meq/kg over 30 minutes, then 2.5 meq/kg/day until serum bicarbonate is 24-28 meq/L HD = last resort but may be lifesaving in severe refractory cases (especially neonates)



Seizures (when due to IEM)

Etiology	Alteration of intracellular **osmolality**, depletion of substrates needed for **cellular metabolism** or **membrane function**, and/or intracellular accumulation of **toxic substances**
DDx	DDx of 'seizures in a newborn' is large, including many IEMs with poor prognosis. **Rare but potentially treatable etiologies**: **pyridoxine responsive** seizures, **folinic acid responsive** seizures, **serine responsive** 3-phosphoglycerate DH deficiency, sz from **hypoglycemia, biotin responsive** holocarboxylase synthetase deficiency, biotinidase deficiency.
Treatment	**See neurology section for treatment of status epilepticus**; avoid AEDs that block mitochondrial fxn (VPA, chloral hydrate) - c/s fosphenytoin, BZDs, and/or levetiracetam. Correct fever, electrolyte issues, acidosis, hypoglycemia. If refractory, c/s empiric pyridoxine (100-200 mg IV x1), folinic acid (2.5-5 mg PO once daily), L-serine (200-600 mg/kg/d div 6x/day), or biotin (5-20 mg PO once daily).