MANCHESTER 1824
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norchi
northern congenital hyperinsulinism service

GDH-HI

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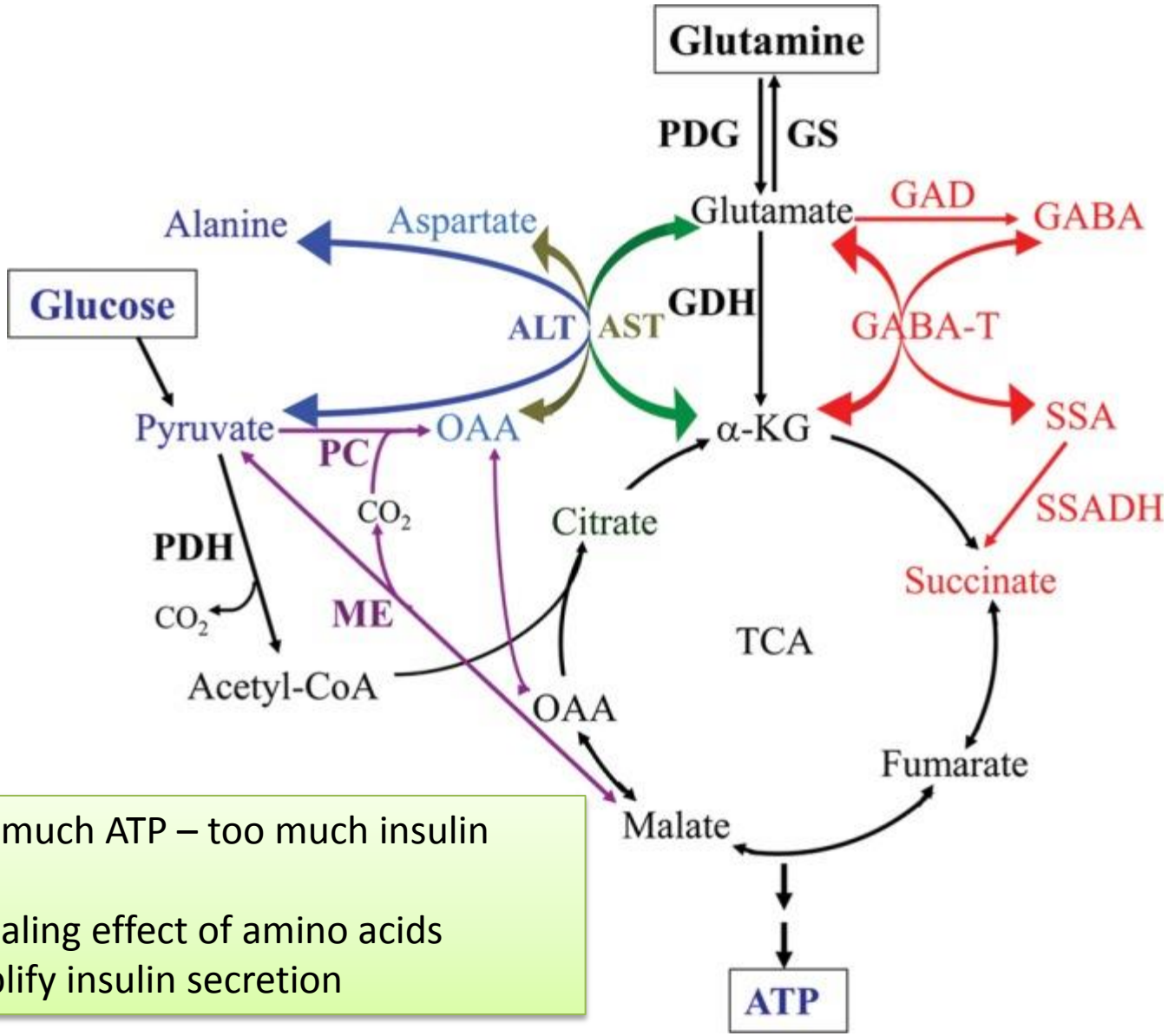

Too much ATP – too much insulin

Signaling effect of amino acids amplify insulin secretion

Presentation

- Late onset
- Low sugars after food
- Fasting hypoglycaemia
- Convulsions, learning difficulties

Adverse Neurodevelopment

- Hypoglycaemia related?
- Direct consequence of GDH enzyme on brain supporting nerve cells (glial cells)? *Komlos et al, Glia 2013*
- Loss of protective neurotransmitter (glutamate) in brain

Patient Stories

- D – 2 year old girl with recurrent hypoglycaemia and seizures
- Ketotic response on at least 2 occasions
- Fast results inconclusive
- Diagnosis of hyperinsulinism when admitted with hypoglycaemic seizures
- Normal ammonia levels

Story of D

- GLUD1 mutation positive
- Diazoxide responsive but erratic adherence
- Social concerns
- Relatively good doses of diazoxide (12 mg/kg/day) but no excess body hair
- Delayed development
- Baby brother with no hypoglycaemia at birth

E’s story

- Diagnosed at age 8 years
- Difficult family circumstances, relationships
- Takes own medication at age 15 years
- Issues with school, home
- Vulnerable young person
- What is life as a young adult

Story of siblings with GDH-HI

- First child (6 years) treated with diazoxide at 15 mg/kg/day – excess hair, worried
- Low protein diet – ProZero milk
- Occasional hypoglycaemia
- What next?
- Second child, age 6 months recently diagnosed with GLUD1 mutation