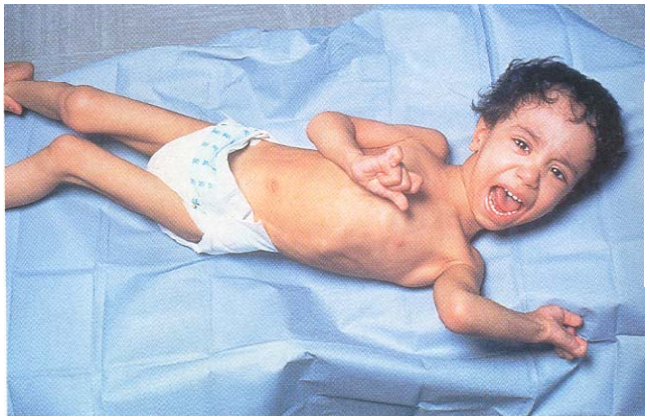
Glutaric acidemia type I:
Clinical features

- Prevalence = ~1/100,000
- No other clinical symptoms at birth- normal development
- Acute encephalopathic episode usually preceded by infection w/ fever and dehydration (2-36 months)
- During episode: profound hypotonia, loss of motor skills, athetoid movements, stiffness, convulsions
- In minority of patients, motor delay, hypotonia, dystonia & dyskinesia develop gradually over several years

OMMBID



Atlas of Metabolic Diseases

Glutaric acidemia type I:
Difficulty in making laboratory diagnosis

- "Low excretors"
  -- Normal or slightly elevated C5-DC in plasma
  -- Normal or slightly elevated excretion of glutaric and/or 3-OH-glutaric acid
  -- Higher residual enzyme activity
  -- Same clinical outcome as "High excretors"
  -- Are patients being missed by NBS and/or follow-up testing?
  -- Urine C5-DC analysis more sensitive?
- Elevated C5-DC observed in other metabolic conditions
- Elevated glutaric and/or 3-OH-glutaric acid excretion caused by intestinal bacteria, severe ketosis or certain infant formulas
- No common mutation in general population (over 100 known)
- No genotype/biochemical phenotype : clinical phenotype correlation

	NBS (<0.32 uM)	plasma (<0.13 uM)
Glutaric aciduria type I		
GAI-1	**0.79**	**0.54**
GAI-2	**0.35**	**0.18**
GAI-3	**0.55**	**0.16**

Glutaric acidemia type I:

- Each patient had normal to mild increases in their urine organic acids
- Either had two mutations or low enzyme activity
- All three are doing well

South Carolina GA1

Patient ID	Original C5DC result with NeoGram >=0.32 uM	C5DC Retest result 2 yrs. Later with NeoGram >=0.32 uM	C5DC Retest result 2 yrs. Later with NeoBase >=0.45 uM
20081901068	0.55	0.32	0.23
20083231218	0.35	0.27	0.25
20083451356	0.79	0.48	0.45