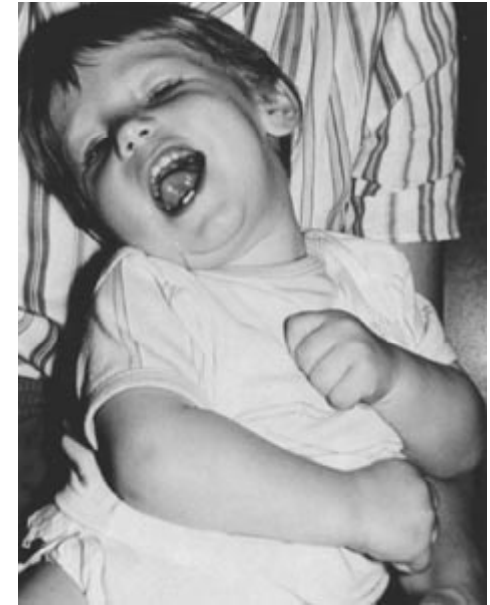


# Glutaric acidemia type I:

## Clinical features

- Prevalence =  $\sim 1/100,000$
- 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

	<b>NBS</b> <b>(&lt;0.32 uM)</b>	<b>plasma</b> <b>(&lt;0.13 uM)</b>
<b>Glutaric aciduria type I</b>		
GAI-1	<b>0.79</b>	<b>0.54</b>
GAI-2	<b>0.35</b>	<b>0.18</b>
GAI-3	<b>0.55</b>	<b>0.16</b>

## 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

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