

# Amino acylase 1 deficiency

Jackie Udall  
Addenbrooke's Hospital

- 5 month old male, presenting with generalised developmental delay and hypotonia
- At 3 years of age, profoundly hypotonic, practically no speech, severe FTT, frequent vomiting

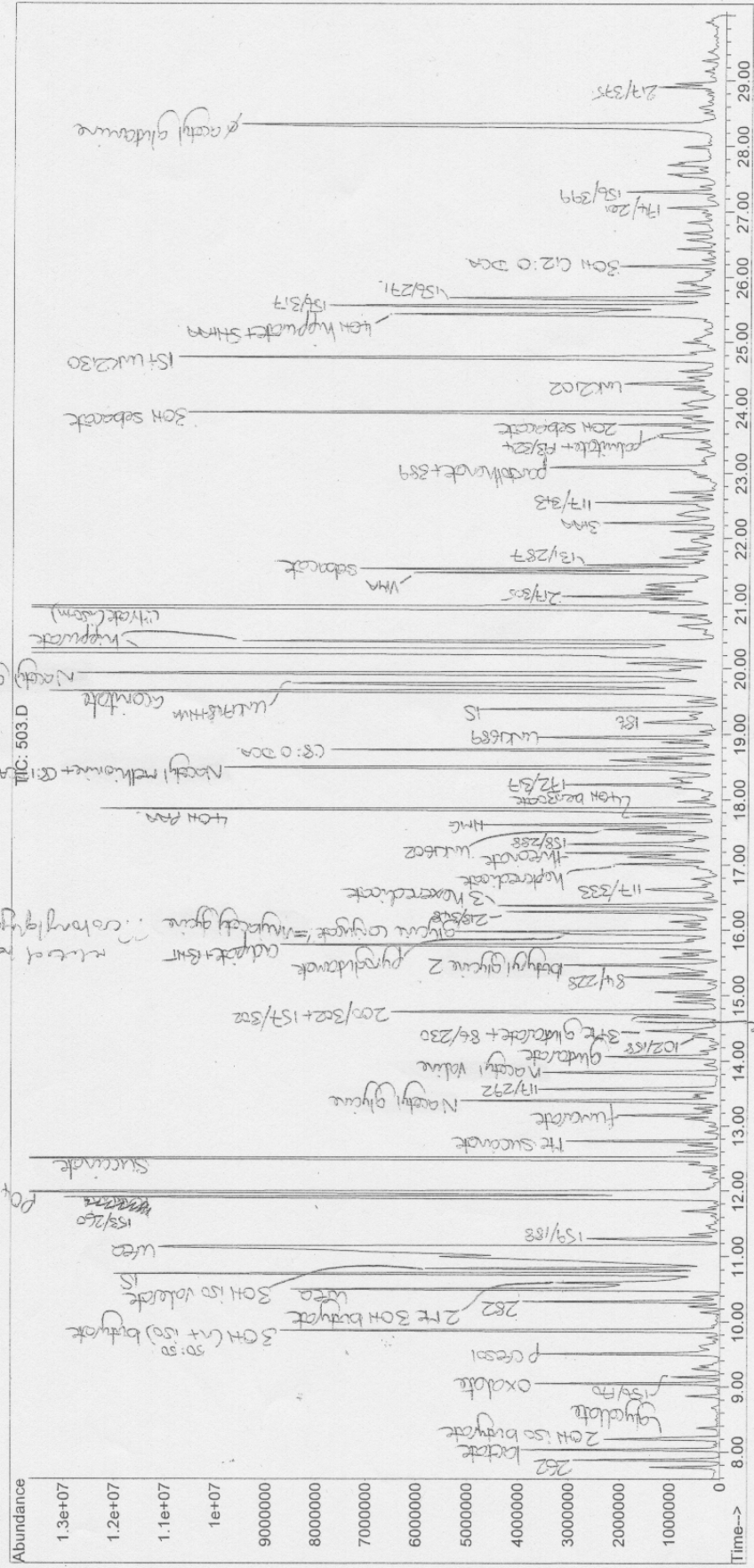
# Results

- Urine organic acids - raised levels of N-acetyl amino acids, butyrylglycine and vinylacetylglycine
- Urine and plasma amino acids - normal
- Urine NMR spectroscopy – raised N-acetyl amino acid levels

# Organic acid profile

File : D:\MSDCHEM\1\DATA\BGU\200505\503.D  
 Operator :  
 Acquired : 13 May 2005 20:40 using AcqMethod BGOA2  
 Instrument : Instrument  
 Sample Name :   
 Misc Info :  
 Vial Number :

158 - N/D  
 102 - 600K  
 40H Butyric  
 30H Glutaric  
 Hexanoyl Glycine 1&2  
 Succinyl Acetone 1&2  
 Orotic Acid  
 Methyl Citrate 1&2  
 butyly glycine detected  
 Suberyl Glycine 1&2



# Has our patient got ACY 1 deficiency?

- Has got the biochemical profile
- Enzyme assay unreliable – deficiency not proven
- No mutations found