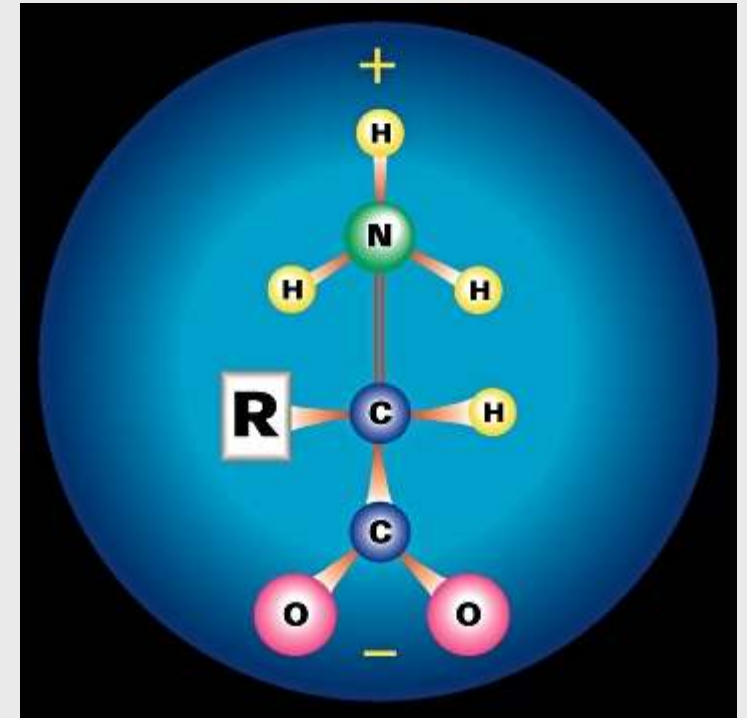


Workshop Introduction

Amino Acids

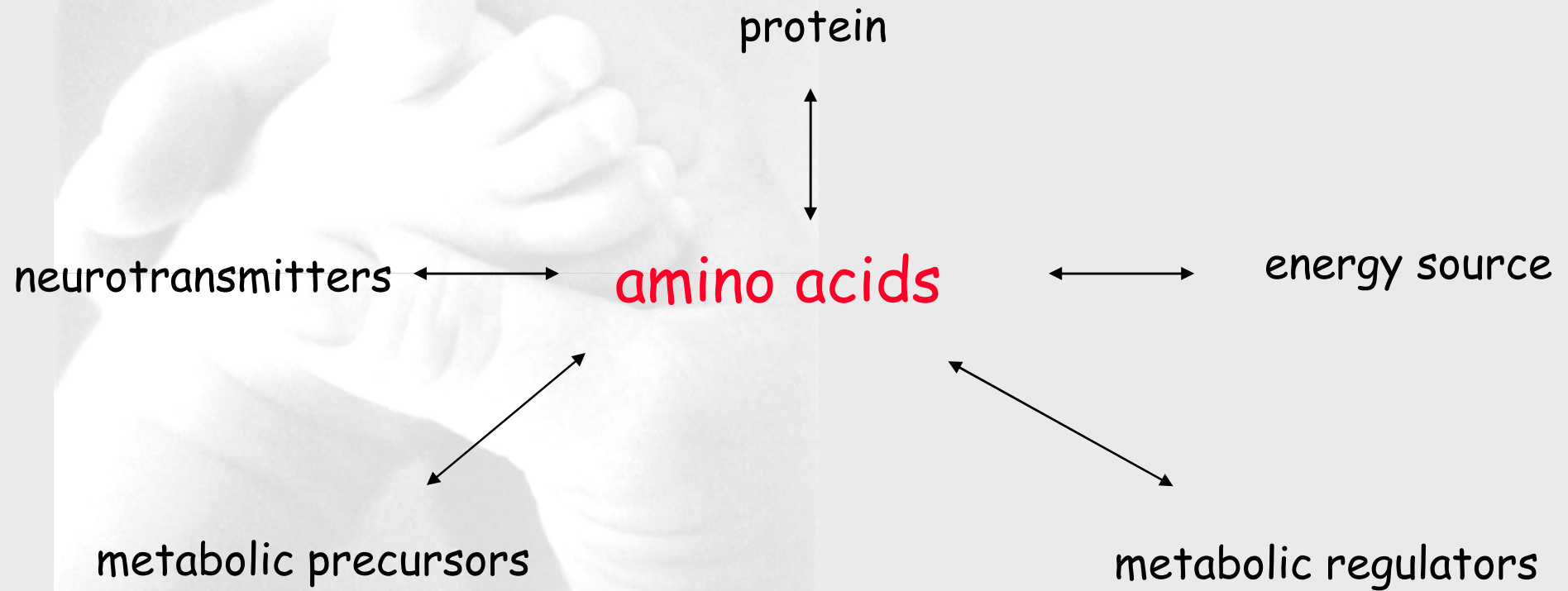
Dr Mick Henderson

Biochemical Genetics
St James's University Hospital, Leeds

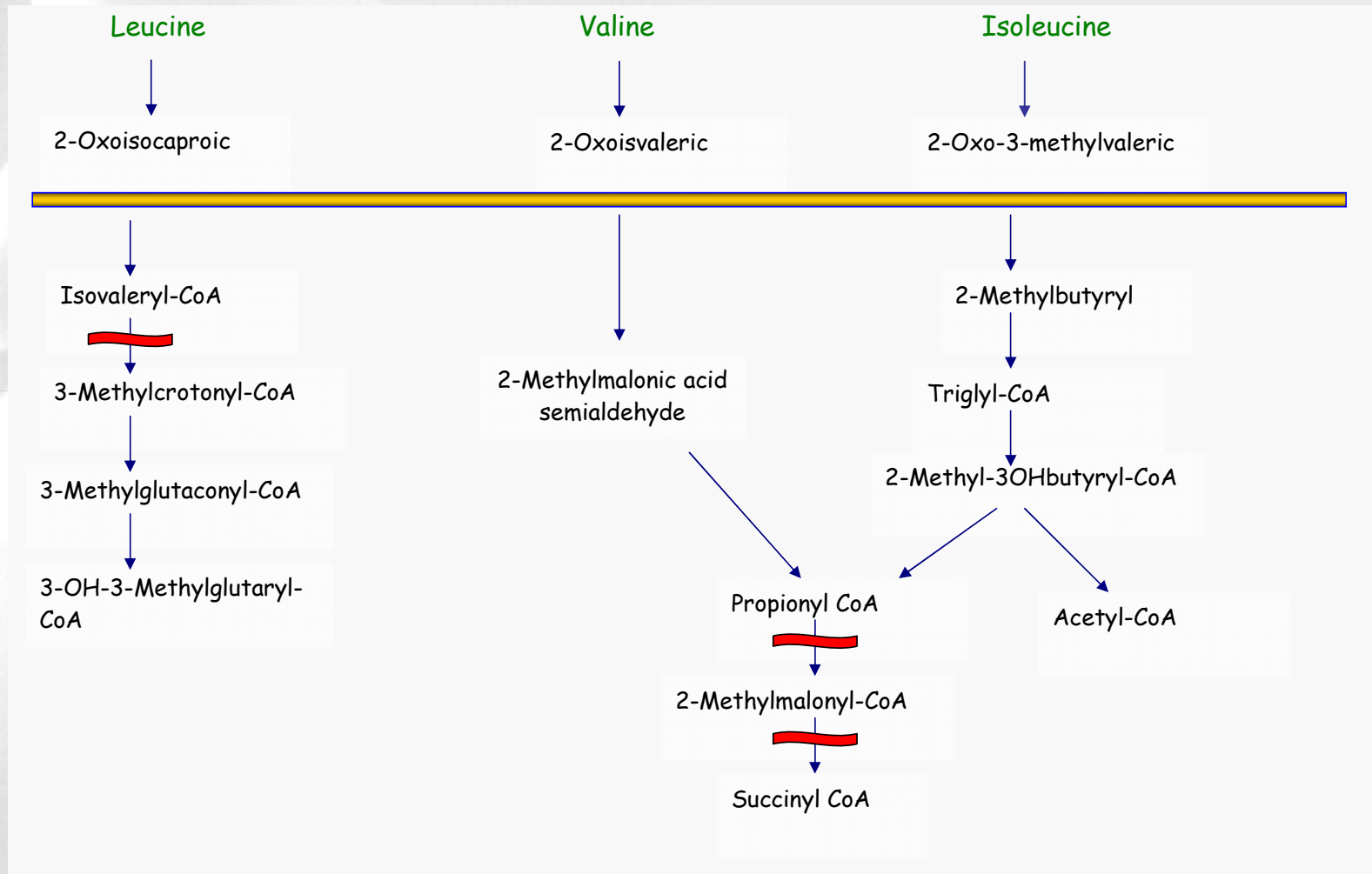


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What use are amino acids?



Branched chain amino acid catabolism



Essential amino acids

Histidine, isoleucine, leucine, lysine, methionine, phenylalanine, threonine, tryptophan, valine

conditionally essential for neonates:

cysteine, tyrosine, taurine, arginine and glycine



Lab methods

- Qualitative
 - TLC, 1D and 2D
 - High voltage electrophoresis
- Quantitative
 - Amino acid analyser - automated ion exchange
 - HPLC
 - UPLC
 - GCMS
 - Tandem mass spectrometry

There is a value to having more than one technique

Which sample type?

Each sample type has advantages and disadvantages

Urine

- Useful for compounds with high renal clearance, e.g. arginino succinate,
- not traumatic to collect

Plasma

- Useful for compounds with low renal clearance, e.g. citrulline
- May be easier to collect than urine

CSF

- Vital for metabolic disturbances confined to CNS, e.g. NKH

Factors influencing interpretation of amino acid results

- **Age**
 - Infancy/feeds
- **Fasting**
- **Time since last meal**
- **Diet**
 - TPN, chicken, etc
- **Infection, ?catabolic**
- **Pregnancy**
- **Specimen quality**
 - Transport
 - Delay in separation of serum
- **Contamination**
 - cells, sweat
- **drugs**
- **Analytical**

Sample Problems

Specimen

- Contamination

ECF, sweat, haemolysis, skin cream, drip fluids.

- Delay

gln → glu; arg → orn; tryp
Loss of cys, hcys

- Storage conditions

Gln → Glu

Other

- Diet
- Drugs
- Bacterial metabolism, effects on urine samples

e.g. carn; ans; hcit; methyl his

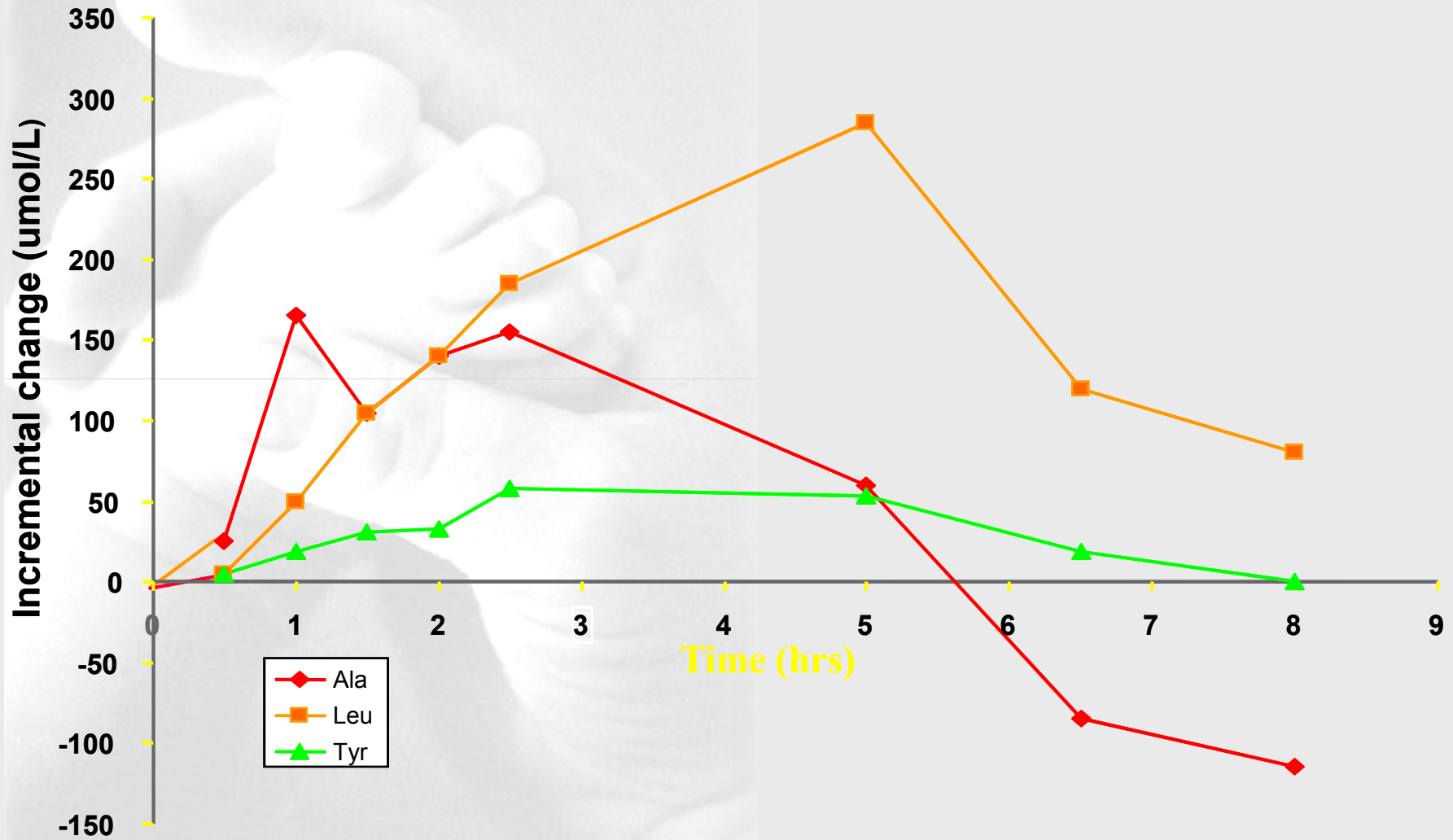
↓cys, ser

Fasting or random samples?

- **Fasting**
 - ideally 6 hrs post intake to reach basal levels
 - reference intervals more reliable
 - dietary artefacts absent
 - milder disorders, or carrier state, may be missed if patient not catabolic

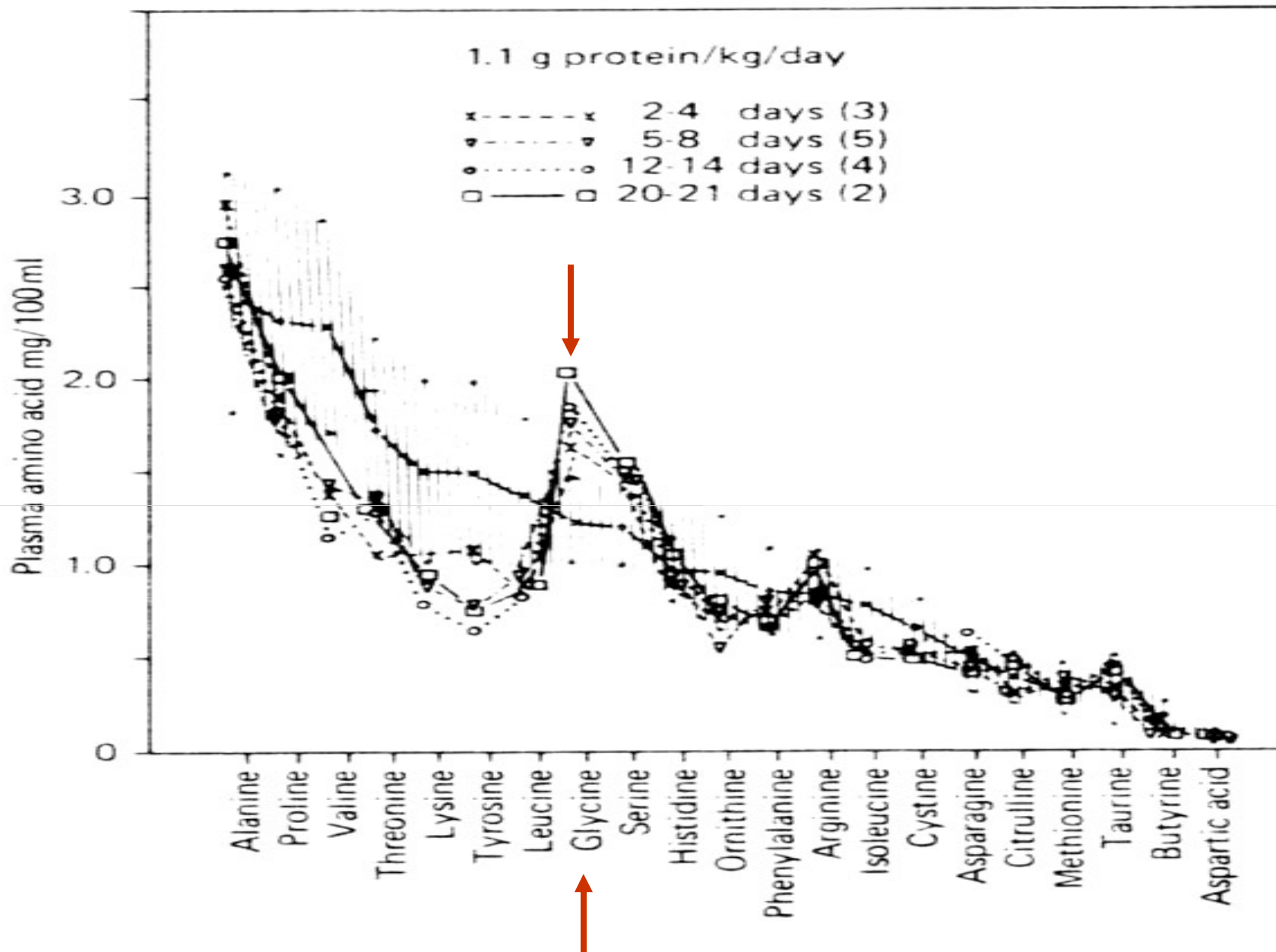
But, fasting often not possible or practical

Changes following 1.2g/Kg protein



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Palmer T et al (1973) *Clin Sci Mol Med* 45: 827-832



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 Snyderman SE, Holt LE et al (1968) *Pediat Res.* 2: 131-144

Drug Interference in Amino Acid Analysis

- Many drugs interfere with amino acid analysis of urine samples
- interference is variable
- Often difficult to obtain accurate drug history in clinical environment
- patients frequently on multiple drug therapy

Red Bulluria!

