Reporting Amino Acids: The Clinical Biochemist

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Why do we measure aminoacids in the laboratory?

- To detect inborn errors of metabolism
- To monitor treatment – PKU, MSUD, argininaemia, ASA, OAT, citullinaemia, cystinuria, tyrosinaemia, homocystinuria
- To assess nutritional wellbeing – TPN, artificial diets
- To identify renal tubular dysfunction
- To investigate renal stones
- To add additional information when investigating hypoglycaemia (alanine), hyperammonaemia (glutamine), lactic acidaemia (alanine)

Inborn errors of metabolism detectable by aminoacid analysis

Phenylketonuria*	Tyrosinaemia types I & II*
Non ketototic hyperglycinaemia*	Maple syrup urine disease
Serine synthesis defects*	Homocystinuria
Cystinuria*	Argininaemia*
Citrullinaemia*	OTC deficiency
Lysinuric protein intolerance*	Hypophosphatasia*
ASA uria*	OAT deficiency*
Hartnup disease*	HHH syndrome*
Prolidase deficiency*	Sulphite oxidase deficiency

*Cases detectable primarily by aminoacid analysis

What kind of turnaround time should we offer?

- Our own experience
  - 50 quantitative aminoacid analyses coming to the lab in July 2005 – Mean turnaround 5.8d, SD 3.0d, range 1-15d
- Detection of IEM's
  - 24h if urgent
  - 10d otherwise
- To monitor treatment
  - 24h if urgent
  - 7d otherwise
- General nutritional assessment
  - 7d
- Renal tubular function
  - 7d
- Renal stones
  - 10d
- As part of the investigation of hyperammonaemia/ hypoglycaemia/ lactic acidaemia
  - 7d

Why is reporting so important?

- It answers a question posed by the requesting clinician
- It acts as a permanent record as part of the patients medical record
- It can be used in future litigation
- It is a serious potential cause of confusion
- It is used to judge the quality of the service by users

What are the features of a good report?

- It is clear, easy to read and unambiguous
- It separates fact from conjecture ie findings from comment
- It contains all necessary information and NO unnecessary information
- It should be suitable for the target audience
- It answers the question and is clear about the next steps if needed
- It is as short as possible

How should we construct an aminoacid report?

- Method used
  - Quantitative or qualitative and very brief method type
  - Source of reference ranges
- Findings
  - If qualitative any particular aminoacids of note
  - Quantitative results with units and age related reference range
- Comment
  - Clinically significant deviation from normal if any
  - Any qualifying concerns eg dilute sample, evidence of sample deterioration or interference

How should we construct an aminoacid report?

- Conclusions
  - No abnormalities detected
  - Nothing diagnostic or No significant abnormality
  - Results requiring further follow-up
  - Possible disorder indicated with a brief differential diagnosis
- Advice for further investigations
  - Repeat or urine/plasma needed
  - Other investigations required
  - Advice about timescale
- Need to test other family members
- Note of whether the result has been telephoned

Common problems

- No sample obtained
- No clinical information provided
- Correct test not requested eg urine homocystine to exclude defects of homocystine metabolism
- Poor sample provided eg too dilute, deterioration, drug interference
- Inadequate analytical reproduciblity eg phe
- Unwillingness to commit to normal
- Lack of explanation about what the abnormal results mean
- Lack of clarity about what to do next and when
- Lengthy reports with too many auto comments
- Results arrive too late
- The wrong person or no-one informed when telephone results are issued

What can we do?

- Ensure a regular dialogue with users, lectures, newsletters, ward rounds and telephone
- Ensure that the clinical question is clearly stated and understood and that relevant clinical details are provided
- Ensure that we have clear written standards for the service that are available (and used!) by staff and users eg clinical details, sample labelling, turnaround time, reporting format, policy on dilute samples, follow-ups etc
- Audit regularly against these standards

What can we do?

- Conduct service evaluation by user questionnaire
- Evaluate whether the service makes a difference
- Modify the service and re-audit at pre-planned intervals. Cost for 2000 samples pa workload. Maybe equipment 20k, reagents & consumables 8k, staff time 50k – Total £78k pa
- Compare practice with other similar centres in UK and Europe
- Explore alternative analytical approaches that may prove more clinically useful and more cost effective

What can we do?

JOIN THE COGNITIVE AMINOACID SCHEME !