Biochemical Genetic Testing in the United Kingdom

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The provision of IMD Services

- Organised from NHS hospitals, birth rate around 740,000 per year, population is 55m
- Three or four larger “Clinical” centres
  - Manchester
  - London
  - Birmingham
  - Cambridge
- A number of others with single handed services
  - Sheffield
  - Cardiff
  - Newcastle
- Some areas without easy access to specialist services
  - Nottingham
  - Bristol

Laboratory testing

- Core tests – most centres
  - organic acids, aminoacids, acylcarnitines, ammonia, intermediary metabolites, MPS, mono/disccaharidases, VLCFA's
- Specialised tests – supra network, some centres (at least two)
  - Lysosomal enzymes, mitochondrial studies inc resp chain enzymes, non-lysosomal enzymes, purine & pyrimidine, other peroxisomal, others
- Screening tests
  - PKU, CHT, CF, Sickle, MCAD

Current provision - core

- Organic acids
  - 13 centres
- Aminoacids
  - 14 centres
- Acyl carnitines
  - 9 centres

Current provision - specialised

- **Lysosomal enzymes**
  - 4 centres
- **Non-lysosomal enzymes**
  - 6 centres with specialisation
- **Purine/pyrimidines**
  - 1 centre
- **Mitochondrial**
  - 3 centres
- **Peroxisomal**
  - 8 centres
- **Galactosaemia etc**
  - 4 centres
- **Porphyrins**
  - 1 centre
- **Molecular Genetics**
  - Around 6 centres, usually as specialised molecular genetic labs some testing in the context of biochemical genetic labs

Current provision - screening

- 16 centres all providing :
  - Cystic fibrosis
  - MCAD
  - Sickle
  - PKU
  - Hypothyroidism
- Typical size, population 3.5m, birth rate 47k pa

MetBioNet

- A DH established 17 laboratory Stakeholder group comprising: Belfast, Birmingham, Bristol (2 labs), Cambridge, Cardiff, Edinburgh, Glasgow, Leeds, Liverpool, London (2 labs), Manchester (2 labs), Newcastle, Sheffield, Southampton.

What did we set out to do?

- Provide a baseline assessment of current services including their scope, capacity, staffing and equipment.
- Perform a risk assessment of the robustness of current services with a view to developing risk sharing and back-up arrangements.
- Assessment of equity of access on a National basis.
- Undertake manpower planning for the specialty.
- Assess national training needs in collaboration with NHS workforce development organisations.
- Provide better information and advice to liaise with accreditation bodies.

What did we set out to do?

- Plan testing for the very rare disorders to ensure National availability.
- Monitor the adequacy of existing EQA schemes and promote development of new schemes as needed.
- Promote best practice guidelines for investigation.
- Conduct/promote clinical audit.
- Promote teaching and education
- Promote and co-ordinate research and developments in this area.

What have we achieved?

- Audit
- Workforce planning and education
- Workshops
- Website
- Quality annual meetings
- Equipment and test inventory
- Rare test planning
- International profile
- All labs accredited

Remaining problems

- National planning is difficult
- Workforce planning is not straightforward
- Rarer assays – enzymology is at risk
- Robustness of some services is poor due to staffing
- Out of hours provision is not always available
- Laboratory accreditation, although very helpful, is rather uninformed
- Clinical provision is patchy, recognition as a specialist service is imminent