

National Metabolic Biochemistry (Biochemical Genetics) Network

Update on Current Activities September 2005

1. Education and Training Strategy

Recent developments include:-

- Training syllabus and logbooks for Specialist HST posts now available
- Case presentations template for web site

The Network is working with NOWGEN to produce a training and educational CD on Medium Chain Acyl Dehydrogenase Deficiency.

2. Workshops

Workshops organised for 2005 are:-

- Amino acid disorders (22nd November, 2005)
- Associate Labs (early 2006)

3. Web Site - www.metbio.net

The metabolic assay directory (MAD) is working well and a disorder search facility is under development - www.metbio.net/directory

The general functionality of the site has been modernised, and now has specific pages on:-

- ♦ Training and Education (www.metbio.net/education)
- ♦ Best Practice Guidelines (www.metbio.net/guidelines)

4. Guidelines currently in preparation are:-

- Unexplained liver dysfunction and metabolic disorders in the older child
- Dysmorphology and inherited metabolic disorders
- Unexplained tubular function and inherited metabolic disorders
- Unexplained neurology in adults with inherited metabolic disorders

5. Quality

The annual Quality Day will take place 18th October 2005.

A review of specialist QA schemes has been completed and is now on the web site (www.metbio.net/quality).

6. Audit

Audit initiatives planned:-

- a re-audit of ammonia measurement is planned for late 2005
- SUDI protocol

7. Service Needs

The Network has completed a report as part of the JCMG National exercise on IMD Service Needs (Hilary Burton), with information and diagnoses across the UK; data on manpower, equipment and workload has also been summarised. A full report is available.

8. Workforce Planning

One of the cluster trainers is to organise workforce data collection across the UK on an annual basis.

8. Publications

The Network will be presenting a poster at the Society for the Study of Inborn Errors of Metabolism annual meeting, 6th - 9th September 2005, in Paris.

The Network has produced a 'flyer' (attached) for circulation to health professionals.

Professor Anne Green
Lead Scientist
National Metabolic Biochemistry Network
September 2005