SSIEM

Training Syllabus

CLINICAL SYLLABUS

SYLLABUS FOR TRAINING IN CLINICAL PAEDIATRIC METABOLIC MEDICINE

Updated July 2006

This syllabus is intended as a guide. Whilst the training should be comprehensive, it is recognised that not all subjects can be covered in the same detail. Changes may also be necessary to meet local needs.

CONTENT

Knowledge	Skills	Attitude	Appendix
Physiology and Biochemistry Paediatric Metabolic Disease Nutrition Genetics Development Ethics	Clinical Technical Management Academic Teaching Organisational Research	Clinical Communication Education Support and leadership Academic/Research Managerial	Inborn Errors Of Metabolism

C.1 KNOWLEDGE

C.1.1 Physiology and Biochemistry

The normal physiology and biochemistry, including changes during childhood, of:

- fluid and electrolyte balance.
- acid - base regulation
- intermediary metabolism including blood glucose and metabolic response to fasting, lactate, ammonia, aminoacids, organic acids and fatty acids.
- oxidative phosphorylation and the respiratory chain
- lipids and lipoproteins
- cholesterol and other sterols
- lysosome and peroxisome metabolism
- purines and pyrimidines
- porphyrins
- calcium metabolism
- bilirubin
- trace metal metabolism
- relevant aspects of brain metabolism including neurotransmitters. The role of the blood brain barrier
- enzyme biochemistry and tissue expression

C.1.2 Paediatric Metabolic Disease

- The pathological and biochemical changes, clinical symptoms, investigations and management in metabolic disorders of these pathways and organelles, as listed in the appendix.

- Drug management. An understanding and experience of the drugs used for the treatment of metabolic disorders.

- Transplantation. The metabolic indications for and long term follow up of
    1. liver transplantation
    2. haematopoietic stem cell transplantation
    3. renal transplantation
- Principles of *ex vivo* and *in vivo* gene transfer and its relevance to IEM

C.1.3 Nutrition

- The normal nutritional requirements of protein, energy, vitamins and minerals for normal growth and development.
- The principles of dietary therapy including the consequences of changes in the intakes of nutrients. Consequences of under-nutrition and specific nutritional deficiencies.

C.1.4 Genetics

- Mechanisms of inheritance and an understanding of molecular genetics including mitochondrial DNA
- Understanding of principles of prenatal diagnosis

C.1.5 Development

- Normal intellectual and psychological development
- Methods of assessment of DQ, IQ, behaviour and neuropsychological function

C.1.6 Research

- Principles of Good Clinical Practice (GCP) in Clinical Trials

C.2. SKILLS

C.2.1 Clinical skills

1. Assessment and investigation of metabolic causes of :

    - Acute encephalopathy
    - Neurological disease to include retardation, regression, fits, movements disorder, myopathy, etc
    - Liver disease including acute liver failure
    - Hepatosplenomegaly
    - Cardiomyopathy
    - Eye disease
    - Nutritional status and growth
    - Dysmorphic patients
    - Skeletal disorders
    - Skin disorders
    - Renal disorders including the Fanconi syndrome

- Muscle disease

2. Emergency management and indications for and the use of:

    - assisted ventilation
    - peritoneal dialysis
    - haemodialysis, haemofiltration and related techniques
    - intracranial pressure monitoring

3. Genetic counselling and counselling prior to prenatal diagnosis

C.2.2 Technical Skills

1. Interpretation of the investigations including understanding analytical, physiological and nutritional factors that influence the results. The use of investigations for long term monitoring of metabolic disorders.

    - Aminoacids
    - Neurotransmitter and biogenic amine metabolites in CSF and other fluids
    - Organic acids (including interpretation of GC/MS data)
    - Ammonia
    - Intermediary metabolites: glucose, FFA, lactate, ketones
    - Carnitines
    - Enzymes studies: specific and non specific assays
    - Glycosaminoglycans
    - Lipids and lipoproteins
    - Peroxisomal enzymes and metabolites (VLCFA, pristanoate, phytanate,etc)
    - Purines and pyrimidines
    - Vitamin, mineral and other nutrient analyses including copper and caeruloplasmin
    - Cholesterol and other sterols
    - Porphyrins, bilirubin and related compounds

2. Indications and interpretation of:
    - fasting test for disorders of intermediary metabolism
    - allopurinol test
    - glucagon test
    - tests for co-factor responsive disorders (such as B12 test for MMA)
    - loading tests (protein, amino acid, MCT,LCT, glucose)
    - screening methods
3. Biopsies -indications for, planning and interpretation of:
    - skin biopsy (for fibroblast culture)
    - liver biopsy
    - muscle biopsy (needle and open)
    - and other biopsies

4. Experience of the use and interpretation of molecular genetic techniques

5. Practical aspects of dietary management:
    - evaluation of nutritional intake and requirements
    - low protein diets and those with controlled aminoacid intake (MSUD, PKU including the management in pregnancy)
    - low and very low fat diets
    - diets for peroxisomal disorders including adrenoleucodystrophy
    - galactose and fructose free diets
    - dietary management of glycogen storage disease
    - emergency /crisis regimens

- electrolyte replacement regimens (for treatment of Fanconi syndrome)

6. Interpretation of psychological and neuropsychometric testing

C.2.3 Management skills

- Understanding of management skills required for development and use of resources in the metabolic unit, including budget control, contracting, strategic planning and writing a business plan.
- Experience of day to day running of a paediatric metabolic service including management of admission policies within the medical directorate system.
- Importance of and involvement in clinical audit
- structure and function of local, regional and national NHS organisation

C.2.4 Academic skills

- Critical evaluation of clinical results from literature review and audit
- Manuscript preparation
- Oral presentation skills

C.2.5 Teaching skills

- Formal and informal teaching skills at undergraduate and postgraduate level
- Development of core teaching material to enable participation in teaching programmes on a regular basis
- Organisation of a postgraduate teaching programme

C.2.6 Organisational skills

- Routine departmental tasks including duty rotas, teaching programmes and postgraduate meetings

C.2.7 Research skills

- Design of clinical trials including medical statistics
- Data organisation and presentation
- Computer literacy

C.3. ATTITUDES

C.3.1 Clinical

- Appreciate the patient and families understanding of metabolic disease
- Understanding of psychological stress of diagnosis of metabolic disorders
- Understanding of ethnic and cultural differences in attitudes to metabolic disorders.
- Understanding of the problems for the patient and family of progressive disorders and disorders in which the natural history is very difficult to predict such as Leigh's disease.
- Working in partnership with others who care for the children to include multidisciplinary teams, education authorities, etc.

C.3.2 Communication

- Effective communication and appropriate approach to parents, children, colleagues GP and community health care staff, research and laboratory staff and managers

C.3.3 Education

- Commitment to continuing self-education and to teaching others

C.3.4 Support and leadership

- Supportive and sympathetic approach to handling families which can be maintained under difficult circumstances
- Understanding and positive approach to the supervision of junior medical staff
- Recognition of and coping with stress in self and others
- Effective leadership of clinical teams

C.3.5 Academic/Research

- Innovative attitude to clinical problems
- supportive to / active in research
- Collaborative attitude to local and national colleagues

C.3.6 Managerial

- Interest in the overall organisation of departmental activities
- Understanding approach to local management structure
- Constructive attitude to the process of decision making
- Acceptance of shared responsibility for the use of resources
- Ability to respond effectively to clinical complaints

APPENDIX

INBORN ERRORS OF METABOLISM

- Disorders of aminoacid and peptide metabolism
- (Phenylketonuria including the management in pregnancy, homocystinuria, MSUD, etc)
- Disorders of organic acid metabolism
- (propionic acidaemia, methylmalonic acidaemia, etc)
- Hyperammonaemia and urea cycle disorders
- Disorders of carbohydrate metabolism
- (Glycogen storage disease, galactosaemia, fructosaemia, etc)
- Disorders of fatty acid oxidation (MCAD,LCHAD, etc)
- Disorders of ketone body metabolism
- Lysosomal storage disorders (MPS, etc)
- Disorders of lipoproteins and lipid metabolism
- Peroxisomal disorders (Adrenoleucodystrophy, Zellweger's syndrome, etc)
- Disorders of purine and pyrimidine metabolism
- Disorders of calcium metabolism
- Disorders of metal metabolism (Wilson's disease, Menkes syndrome,etc)
- Congenital lactic acidoses and metabolic myopathies
- Porphyrias
- Disorders of cholesterol, sterol and bile acid metabolism
- Disorders of vitamin metabolism (biotin, cobalamin,etc)
- Defects of membrane transport (lysinuric protein intolerance, etc)
- Defects of glycosylation
- Defects of connective tissue