

Syllabus, Curriculum or Logbook/Portfolio?

- Syllabus
 - A list of the topics to be covered in a course
- Curriculum
 - Information on the relative importance of the topics, how they are to be learnt, in what order they are to be learnt, where they are to be learnt and how knowledge is to be assessed
- Logbook/Portfolio
 - A book that that contains information from the syllabus that can be filled in by the student (with possible authorisation by a trainer) to assess the extent and level of their training and highlight further needs.

Available Syllabuses for IMD Training

- **ACB syllabus for Grade A trainees,**
 - Small component of IMD
- **SSIEM Syllabus for Laboratory IMD**
 - Syllabus does not go into a lot of depth and covers areas already covered in BSc, ACB & MRCPPath training eg Basic Biochemistry, Basic pipetting skills, management etc.
- **BIMDG Website**
 - Syllabus aimed at Clinical Paediatric Metabolic Medicine. However there is substantial overlap with a putative IMD syllabus so in principal some training components (eg e-learning) could be shared.

Grade A Trainee Syllabus

(Trainees are not expected to have in-depth knowledge of all inherited metabolic defects but should be aware of the major categories of defect; how they present and are investigated; mechanisms of inheritance; the principles of treatment; and the scope of prenatal diagnosis.)

Quantitative and qualitative enzyme abnormalities occurring in genetic disorders

The biochemical consequences of a primary enzyme block in a metabolic pathway and the way in which clinical and pathological signs may be produced

Methods of detecting metabolic disorders with particular consideration to:

1. • screening selected clinical groups
2. • evaluation of detection programmes
3. • prenatal diagnosis

Methods of treatment, particularly by:
dietary manipulation

Biochemical monitoring of treatment

Amino acid disorders especially those involving:

1. • phenylalanine
2. • tyrosine
3. • branch chain amino acids and maple syrup urine disease
4. • methionine and homocystine
5. • the transport disorders, cystinosis, cystinuria and Hartnup disease

Carbohydrate disorders including:

1. • glycogen storage diseases
2. • galactosaemia
3. • hereditary fructose intolerance and essential fructosuria

Urea cycle defects

Organic acid disorders

Lysosomal storage disorders

Mitochondrial disorders

Peroxisomal disorders

Purine and pyrimidine disorders (including primary and secondary hyperuricaemias)

Cystic fibrosis

Grade A Trainee Syllabus –overlap topics

- Hypoglycaemia
- Hyperammonaemia
- Hyperlipidaemia
- Lactic Acidosis
- Muscle Disease
- Liver Disease

HST Training Syllabus

- Paediatric Biochemistry
- Newborn Screening
- Inherited Metabolic Disease

HST Training Syllabus

- Knowledge of Metabolic Diseases
- Knowledge and experience of analytical techniques and tests.
- Diagnostic problem solving of patients with possible inherited metabolic disease.

HST Training Syllabus

- Need to define:-
 - Core Components
 - Optional components
- Need to consider what happens if an HST leaves the post before completing the course but continues working in an IMD laboratory.