Subject Knowledge Areas

01. Normal Structure and Function
   01.1. Metabolic Pathways
   01.2. Cell Biology

02. Classification of Abnormalities/Pathogenesis
   02.1. Enzymopathies
   02.2. Receptor, transporter, structural
   02.3. Disorders of development
   02.4. Cell biology/structure
   02.5. Mitochondrial disorders
   02.6. Contiguous gene syndromes
   02.7. Peroxisomal
   02.8. Lysosomal
   02.9. Nutritional
   02.10. Other

03. Laboratory Techniques
   03.1. Enzyme assays
   03.2. Analyte
   03.3. Newborn Screening

04. Clinical Management of an individual
   04.1. Diet
   04.2. Drugs/pharmacogenetics
   04.3. Enzyme replacement/gene therapy
   04.4. Bone marrow transplantation
   04.5. Organ transplantation

05. Quality Management
   05.1. Quality Assurance
   05.2. Quality control
   05.3. Quality Improvement
   05.4. Systems Management

Tasks: Application of Knowledge

01. Diagnosis of patient
   T01.1. Techniques
      T01.1.1. HPLC/amino acids
      T01.1.2. GCMS/organic acids
      T01.1.3. TMS/carnitine
      T01.1.4. Enzymes
      T01.1.5. Other

   T01.2. Use of data
02. Management

T02.1. Treatment
   T02.1.1. Symptomatic
   T02.1.2. Presymptomatic

T02.2. Surveillance

T02.3. Counseling

T02.4. Laboratory