Newborn Screening & Detection of Inborn Errors of Metabolism

Content Outline

1. Definition & Classification of Inborn Errors of Metabolism (IEM)
   1.1 General signs & symptoms manifesting in the newborn period
   1.2 Unique physical, laboratory & diagnostic findings in major IEM examples
   1.3 Emergency approach to general stabilization pending diagnosis

2. Newborn Screening (NBS)
   2.1 Purpose, goals & evolution of NBS
   2.2 Recommended Uniform Screening Panel (RUSP)
      2.2.1 Core disorders & recommended secondary disorders in RUSP
   2.3 Screening procedure
      2.3.1 Specimen collection
      2.3.2 Timing of collection & impact of special circumstances
      2.3.3 Reporting abnormal results
      2.3.4 Overview of screening marker distribution curves
      2.3.5 Causes of false-positive results
   2.4 Specific examples of metabolic diseases by category
   2.5 Other non-metabolic disorders in NBS
      2.5.1 Hearing
      2.5.2 Sickle cell disease
      2.5.3 Cystic fibrosis
      2.5.4 Critical congenital heart disease

Reading Material Resources

Module WB2001: Newborn Screening & Detection of Inborn Errors of Metabolism is based on the resources listed below. A copy of each resource is included with the module.

Chapter 21 Introduction to Metabolic and Biochemical Genetic Diseases, Cederbaum S in Avery’s Diseases of the Newborn, 10th ed. (2018), Elsevier, 224-229