Established and Emerging Treatments for Patients With INBORN ERRORS OF METABOLISM

Three Main Approaches to Management

1. Decreasing Substrate
   - Substrate Reduction Therapy (eg, in HT-1)
   - Dietary Restriction (eg, in PKU)
   - Scavenger therapy (eg, in Urea Cycle Disorders: Na benzoate, Na phenylbutyrate, glycerol phenylbutyrate)

2. Provide Product That Is Deficient
   - Examples: Tyrosine in PKU, Arginine or Citrulline in Urea Cycle Disorders, MCTs or Triheptanoin in VLCAD

3. Enhance Conversion of Substrate to Product
   - Liver Transplantation and Human Hepatogastrointestinal Liver Cell Transplantation (eg, in OTC deficiency and Maple Syrup Urine disease)
   - Gene Therapy: Using adenosomal and lentiviral vectors

Sources:
- American Academy of Pediatrics

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