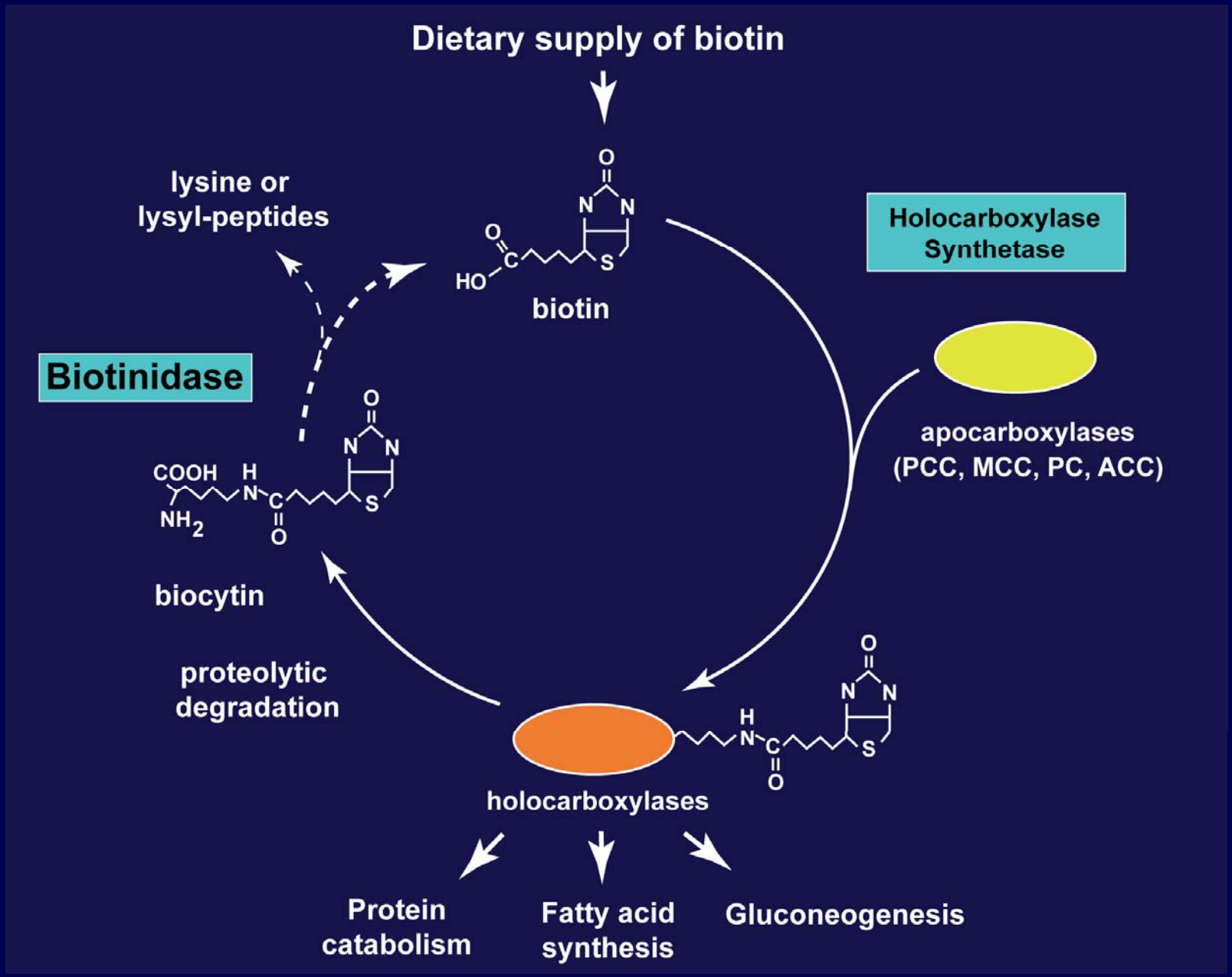
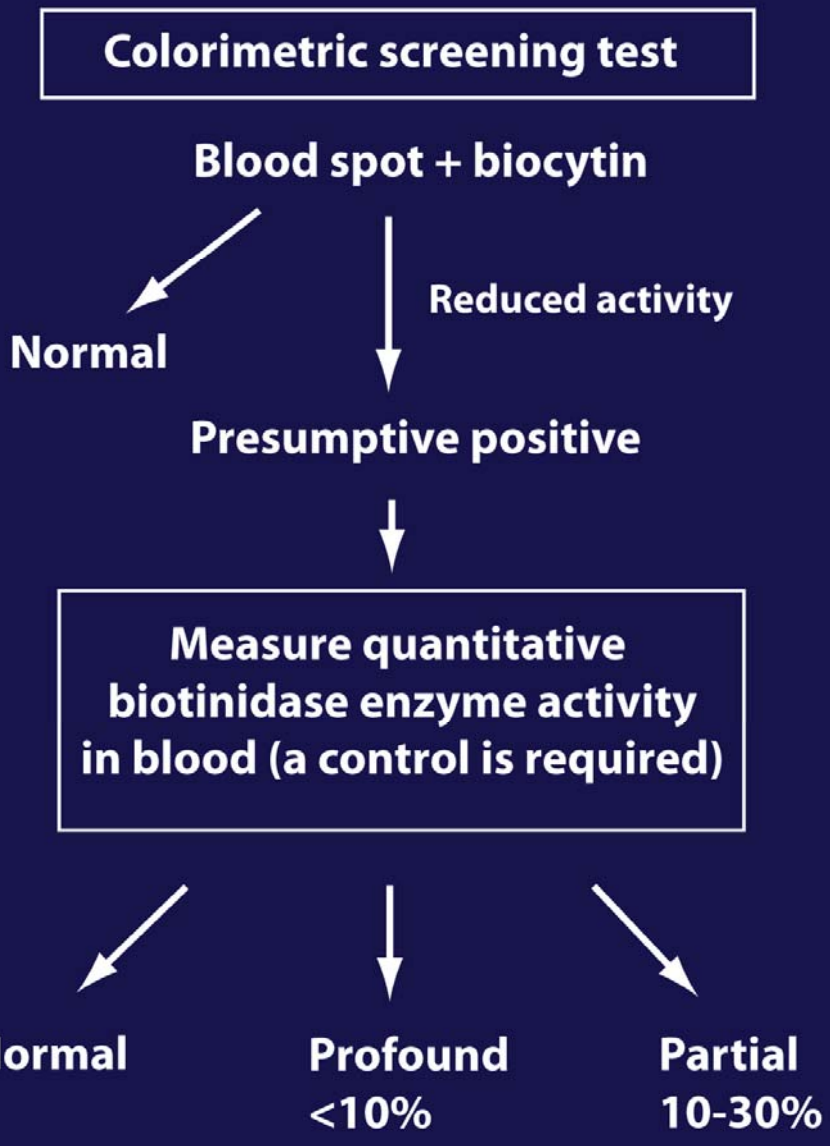
Biotinidase Deficiency

- Impairment of:
  - Fatty acid and glucose synthesis
  - Amino acid catabolism
- Untreated: seizures, hypotonia, ataxia
- Two types:
  - Profound <10%
  - Partial 10% - 30%
- Incidence of 1/61,000 births
- Autosomal recessive
  - Parents are carriers
  - 25% recurrence risk


Dietary supply of biotin
lysine or
lysyl-peptides
biotin
Holocarboxylase
Synthetase
Biotinidase
apocarboxylases
(PCC, MCC, PC, ACC)
biocytin
proteolytic
degradation
holocarboxylases
Protein
catabolism
Fatty acid
synthesis
Gluconeogenesis

Newborn Screening Approach



Treatment

- Profound deficiency
  - 5-10 mg/day oral biotin supplement
- Partial: controversial
  - 5-10 mg/day biotin
  - Supplement with biotin only during illness or stress
- Carriers do not need treatment