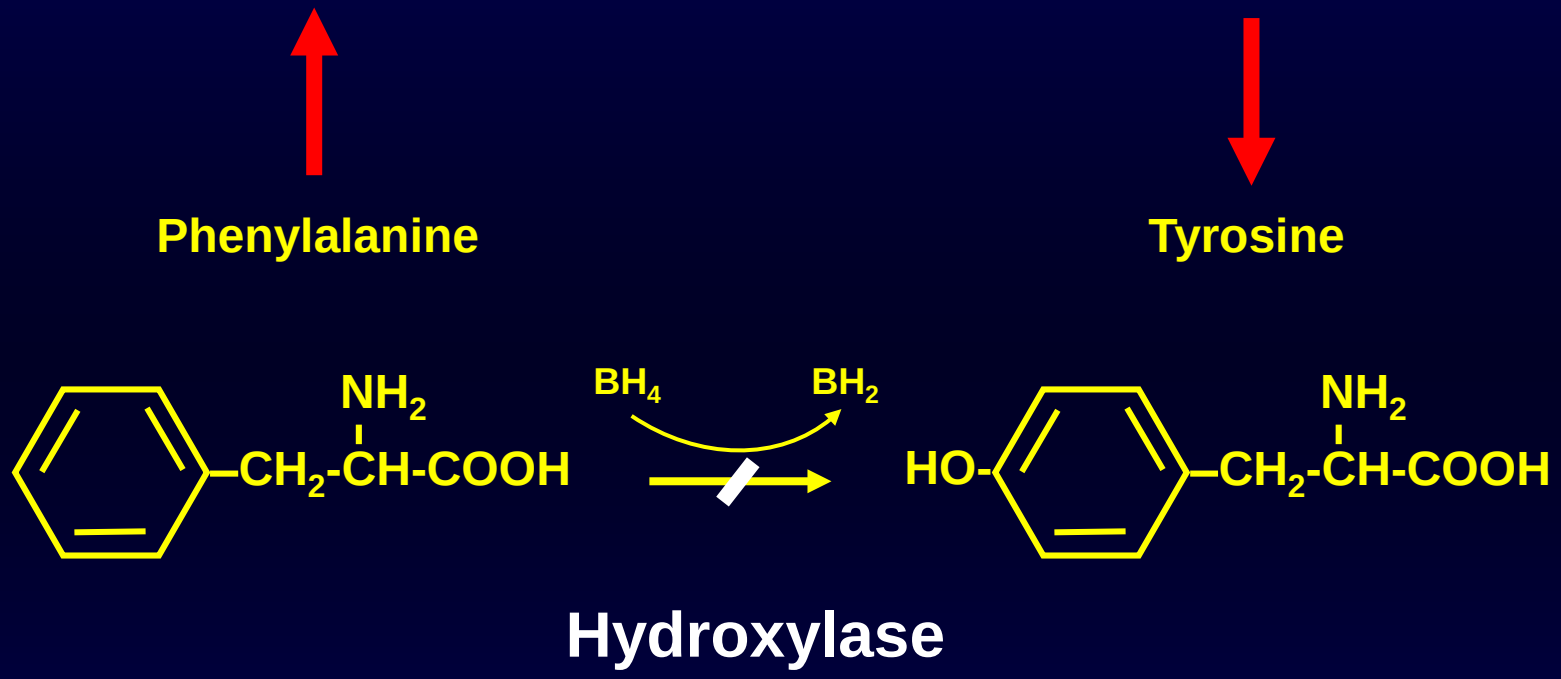
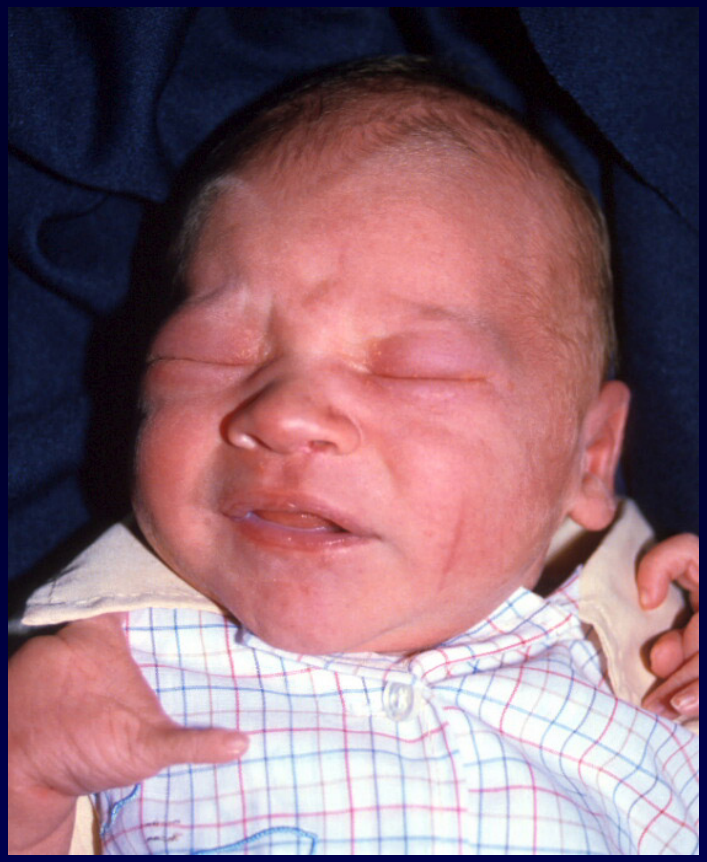
Phenylketonuria

- Autosomal recessive: 1/15,000
- Inability to metabolize phenylalanine
- Most mutations are in hydroxylase enzyme
- ≥ 2.5 mg % is a presumptive abnormal
- Treated by dietary restriction
- Intellectual deficiency if untreated

Metabolism of Phenylalanine



No abnormal clinical findings in PKU newborns



Treatment

- Confirm Diagnosis by age 2 weeks
- Reduce levels to < 6 mg % by age 3 weeks
- Weekly then monthly levels, first 3 years
- Lifelong restricted diet. Developmental outcome is expected to be normal.
- Family planning for affected females
  - Phenylalanine is a potent teratogen

Maternal PKU Fetopathy

- Maternal PHE Levels > 10m%: high frequency of:
  - microcephaly
  - congenital heart disease
  - nonspecific facial dysmorphism
  - most have developmental delay
- Affected children usually have normal enzyme activity