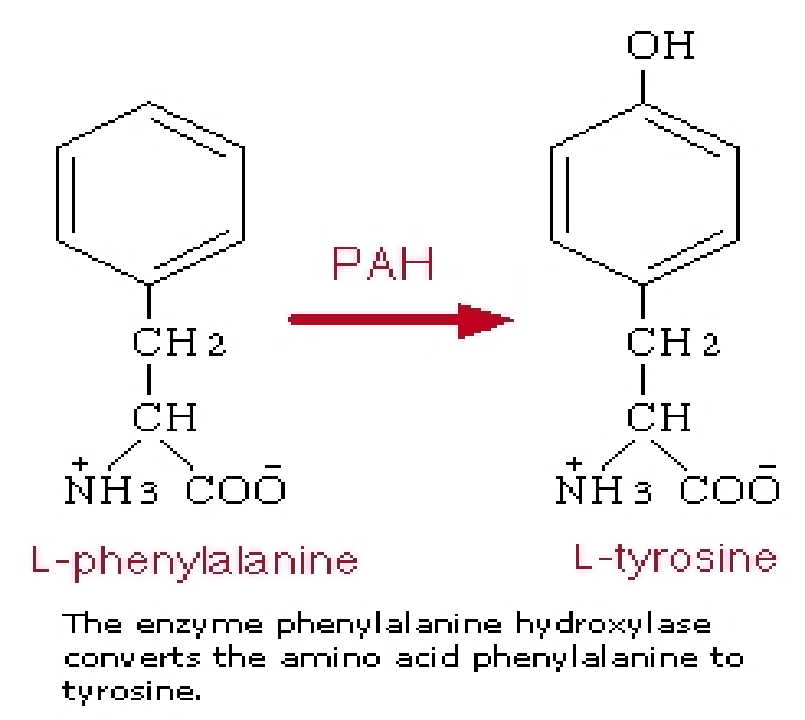
Phenylketonuria

Laurel Fuentes

BIOC 118Q

PKU



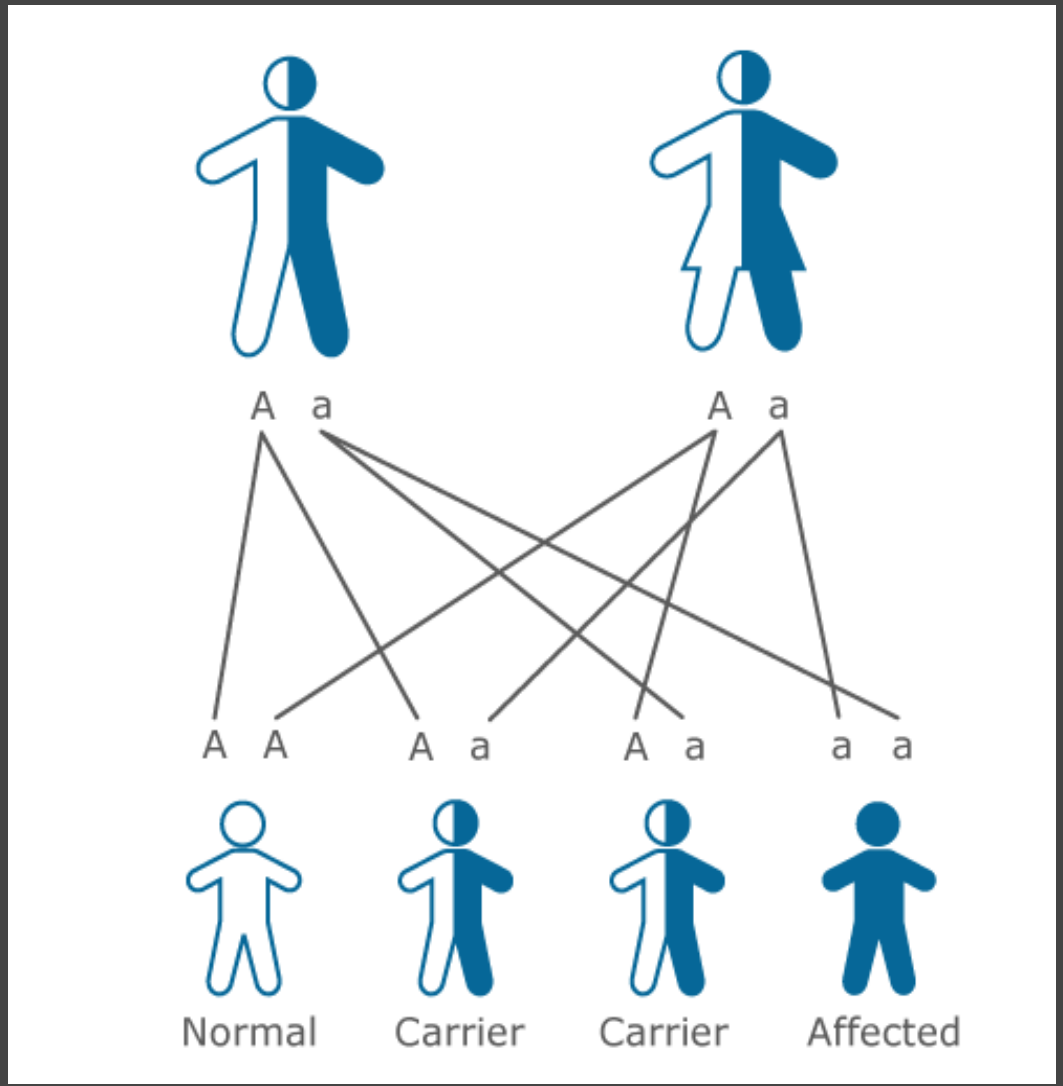
- Occurs in 1 in 12,000 in North America
- Inherited metabolic disease
  - Caused by a deficiency in the enzyme phenylalanine hydroxylase (PAH)

- Symptoms:

mental retardation, organ damage, unusual posture and can severely compromise pregnancy

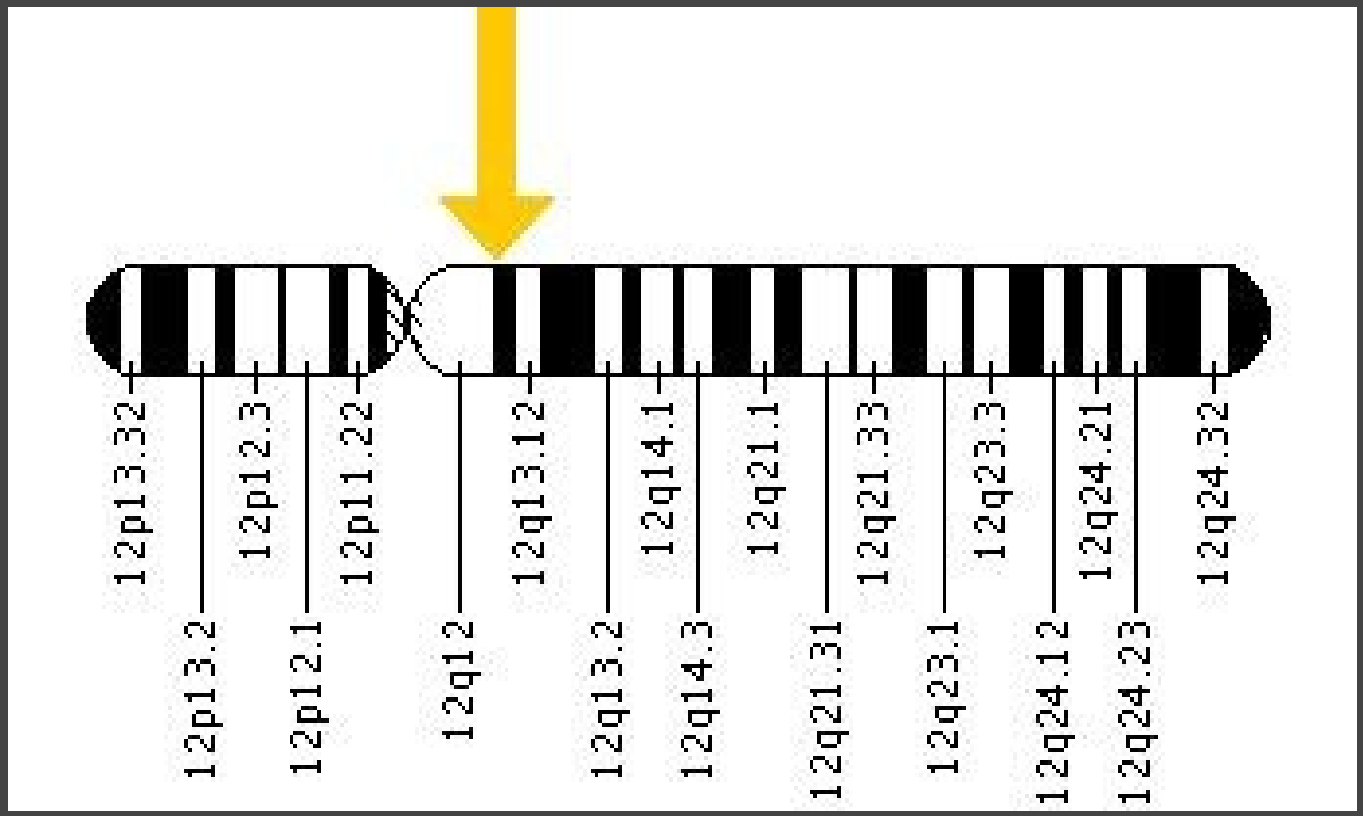
Inborn Errors of Metabolism

- Archibald Garrod - 1908
- PKU - Autosomal recessive disease



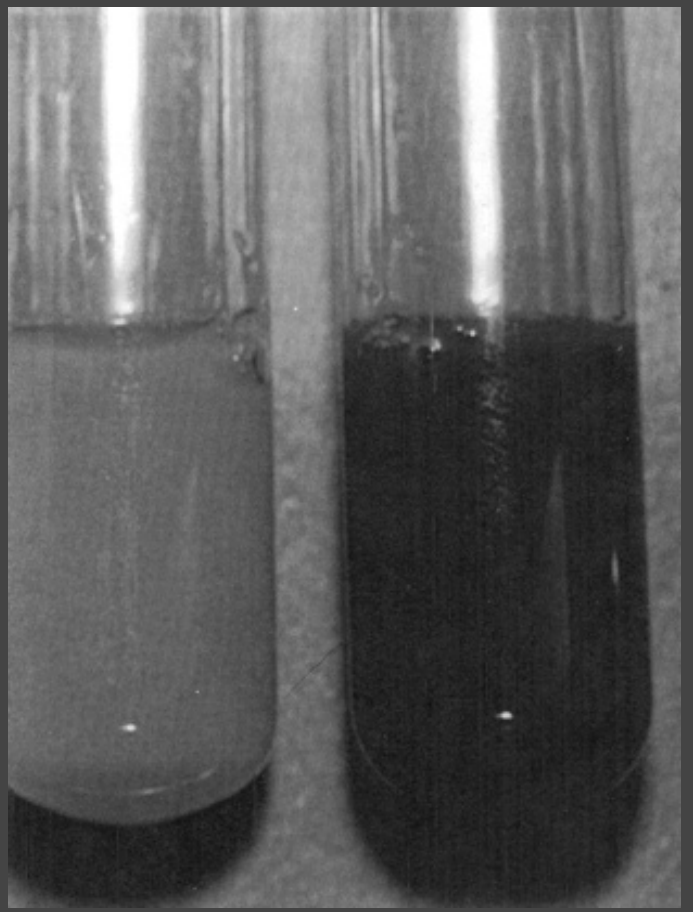
Genetics

- PAH enzyme coding region on chromosome 12 in the bands 12q22-24
- Base pairs on Chromosome 12: 103,232,103 to 103,311,380
- 500 mutations



Classical Diagnosis

- First discovered by Norwegian Doctor, Asbjörn Fölling 1934
- Diagnosis needed the use of multiple pedigrees

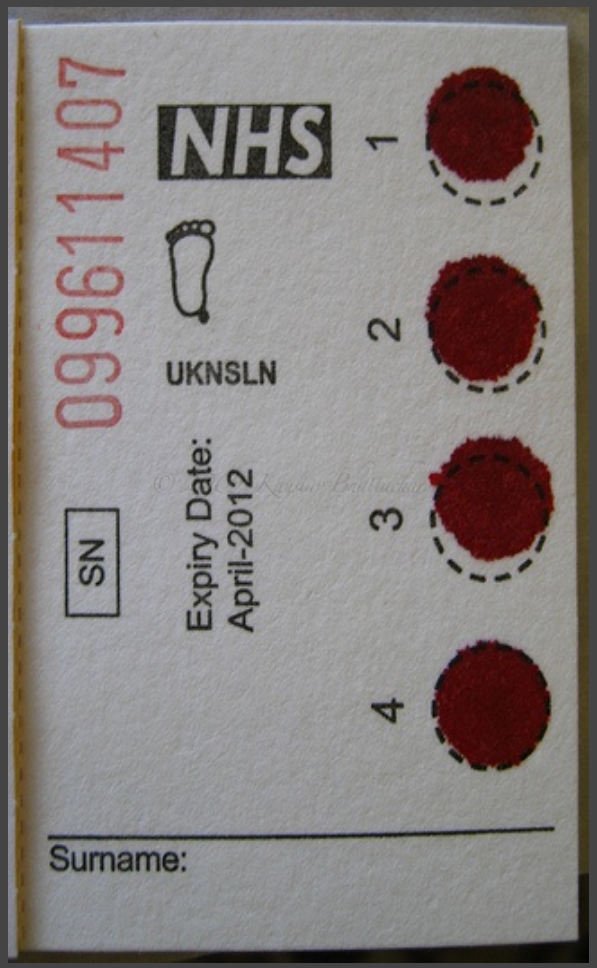


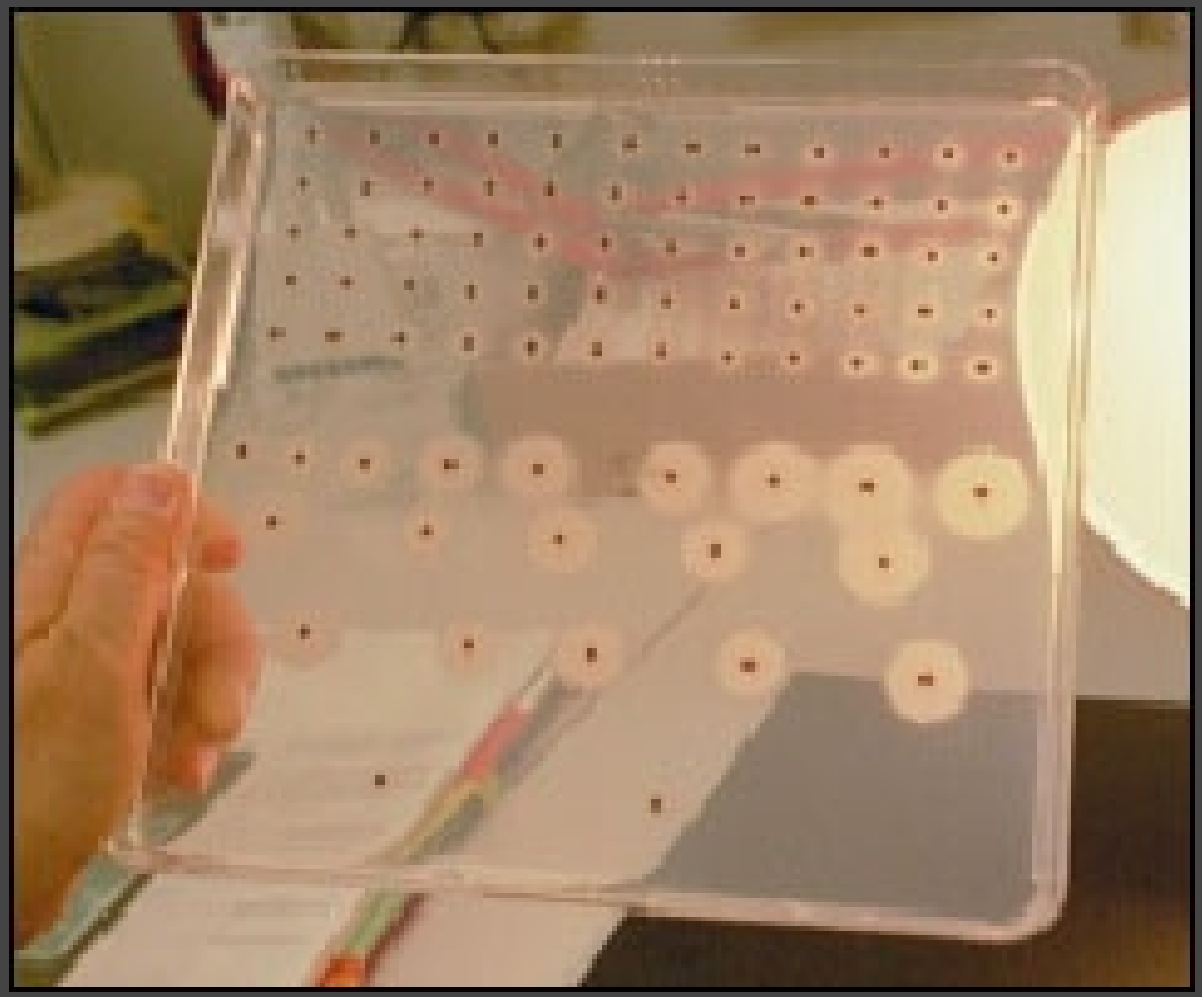
Classical Treatment

- Until 1950s - nothing could be done
- 1950s - special diet food, ferric chloride test

Novel Diagnosis

- Guthrie Test





Novel Diagnosis

- Tandem Mass Spectrometry

Novel Treatment

- No treatment if not caught early
- Preclinical Research:
  - New Gene Therapy Cures PKU in mice

Works Cited

Chu, Wai Lang. "New Gene Therapy Technique cures PKU." *Outsourcing-Pharma*. 11 Oct 2005.

"PAH phenylalanine hydroxylase." *NCBI Entrez Gene*. 12 Jan 2012.

"Phenylketonuria." *OMIM. 27 Man 2011.*

*"*Phenylketonuria." *NCBI Genes and Diseases.* 2008

"Tandem mass spectrometry in newborn screening." *ACMG/ASHG*. **2**. 4. August 2000.