

Disease

# Phenylketonuria<sup>1, 2</sup>

Gene

## PAH

|                                                 |                                                                                                                                                                                                                                                 |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                              |
| <b>Current Treatments</b>                       | low protein diet, pegvaliase, tetrahydrobiopterin                                                                                                                                                                                               |
| <b>Evidence for Treatment</b>                   | guideline <sup>3, 4</sup>                                                                                                                                                                                                                       |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                             |
| <b>Non-Molecular Test</b>                       | Plasma amino acid analysis <sup>1</sup>                                                                                                                                                                                                         |
| <b>ClinGen</b>                                  | <a href="https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC110">https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC110</a>                                                                       |
| <b>Disease Frequency (per 100,000)</b>          | 6.6 <sup>5</sup>                                                                                                                                                                                                                                |
| <b>Genes Included in Disease Frequency</b>      | PAH                                                                                                                                                                                                                                             |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Phenylketonurias&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Phenylketonurias&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1504>
- <sup>2</sup> <https://www.sciencedirect.com/science/article/pii/S1096719220301451>
- <sup>3</sup> [https://www.acmg.net/docs/ACMG\\_PAHPKU\\_Guidelines\\_GIM\\_Feb2014.pdf](https://www.acmg.net/docs/ACMG_PAHPKU_Guidelines_GIM_Feb2014.pdf)
- <sup>4</sup> <https://www.ncbi.nlm.nih.gov/pubmed/29025426>
- <sup>5</sup> <https://pubmed.ncbi.nlm.nih.gov/25085281/>