

Title: Tyrosinemia Type I *GeneReview* — Table 6

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

**Table 6. *FAH* Pathogenic Variants Identified in North American Patients with Tyrosinemia Type I**

| DNA Nucleotide Change<br>(Alias <sup>1</sup> ) | Predicted Protein Change | Reference Sequences         |
|------------------------------------------------|--------------------------|-----------------------------|
| c.1A>G                                         |                          | <a href="#">NM_000137.2</a> |
| c.81+2T>G<br>(IVS1+2T<G)                       |                          |                             |
| c.81+38insG<br>(IVS1+38insG)                   |                          |                             |
| c.192+1G>T<br>(IVS2+1G>T)                      |                          |                             |
| c.193-14G>A<br>(IVS2-14G>A)                    |                          |                             |
| c.257A>C                                       | p.Gln86Pro               |                             |
| c.314-14G>A<br>(IVS3-14G>A)                    |                          |                             |
| c.398G>T                                       | p.His133Leu              |                             |
| c.424A>G                                       | p.Arg142Gly              |                             |
| c.455G>A                                       | p.Trp152Ter              |                             |
| c.459G>A                                       | p.Ter152Ter              |                             |
| c.520C>T                                       | p.Arg174Ter              |                             |
| c.536A>G                                       | p.Glu174Arg              |                             |
| c.542_553+4ins                                 |                          |                             |
| c.548_554+20del                                |                          |                             |
| c.554-1G>T                                     |                          |                             |

| DNA Nucleotide Change<br>(Alias <sup>1</sup> ) | Predicted Protein Change | Reference Sequences         |
|------------------------------------------------|--------------------------|-----------------------------|
| (IVS6-1G>T)                                    |                          |                             |
| c.606T>C                                       | Met202Ile                | <a href="#">NM_000137.2</a> |
| c.607-6T>G<br>(IVS7-6T>G)                      |                          |                             |
| c.620A>G                                       | p.Gly207Asp              |                             |
| c.709C>T                                       | p.Arg237Ter              |                             |
| c.710G>A                                       | p.Arg237Gln              |                             |
| c.782C>T                                       | p.Pro261Leu              |                             |
| c.786G>A                                       | p.Trp262Ter              |                             |
| c.1009G>A                                      | p.Gly337Ser              |                             |
| c.1020C>T                                      | p.Arg341Trp              |                             |
| c.1025C>T                                      | p.Pro342Leu              |                             |
| c.1027C>T                                      | p.Gly343Trp              |                             |
| c.1062+5G>A<br>(IVS12+5G>A)                    |                          |                             |
| c.1069G>T                                      | p.Glu357Ter              |                             |
| c.1090G>T                                      | p.Glu364Ter              |                             |
| c.1116_1123del                                 |                          |                             |
| c.1298_1309in                                  |                          |                             |

1. Variant designation that does not conform to current naming conventions