

Supplemental Data

Molecular Expression and Characterization of Erythroid-Specific 5-Aminolevulinate Synthase Gain-of-Function Mutations Causing X-Linked Protoporphyrria

David F Bishop, Vassili Tchaikovskii, Irina Nazarenko, and Robert J Desnick

Online address: <http://www.molmed.org>

The Feinstein Institute
for Medical Research North Shore LIJ

Supplementary Table S1. Oligonucleotide primers for mutagenesis and sequencing of ALAS2.

| Mutation | Primer ^a | Sequence | Plasmid Location ^b |
|-----------|---------------------|-------------------------------------|-------------------------------|
| delAT | 1F | GTACACTTGAGCTCGAGTGAGTGGAACG | 4138 to 4169 |
| | 1R | CGTTCCCACTCACTCGAGCTCAAAGTGTAC | 4138 to 4169 |
| delAGTG | 2F | GTACACTTGAGCTCATGAGTGGAACGTTCTAC | 4138 to 4176 |
| | 2R | GTTAGAACGTTCCCACTCATGAGCTCAAAGTGTAC | 4138 to 4176 |
| delG | 3F | CTACTTCGGAACATGGGCCAGTATGTC | 4173 to 4203 |
| | 3R | GACATACTGGGCCATGTTCCCGAAGTAG | 4173 to 4203 |
| 548X | 4F | GGGGCTGCCCTCTAGGATGTGTCTGT | 4083 to 4410 |
| | 4R | CACAGACACATCCTAGAGGGGCAGCCCC | 4083 to 4410 |
| 581X | 5F | GGAACATGGGCCCTAGTATGTCACCACC | 4181 to 4209 |
| | 5R | GGTGGTGACATACTAGGGCCCCATGTTCC | 4181 to 4209 |
| Wild type | 6F | TGTTGCCAATGACTCTACTC | 3231 to 3251 |
| Vector | 7R | GTGCCAAGCTTGCCTGCA | 4402 to 4419 |
| Wild type | 8R | AATTGCAGGCAGCCACAGA | 4105 to 4123 |

^a 'F' denotes Forward primer; 'R' denotes Reverse primer.

^b For plasmid pMALc2-AE2; Asp 79 to C-ter = 2689 to 4215