Title: POLR3-Related Leukodystrophy *GeneReview* Tables 3-5
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

**Table 3. Selected *POLR3A* Pathogenic Variants**

| DNA Nucleotide Change | Predicted Protein Change | Exon | Reference(s) | Reference Sequences |
|---|---|---|---|---|
| c.169G>A | p.Asp57Asn | 2 | 1 | NM_007055.3 NP_008986.2 |
| c.272C>T | p.Pro91Leu | 3 | 1 | |
| c.364_366delAAG | p.Lys122del | 4 | 1 | |
| c.418C>T | p.Arg140Ter | 4 | 2 | |
| c.441dupT | p.Asp148Ter | 4 | 1 | |
| c.496G>A | p.Val166Ile | 5 | 1 | |
| c.550_553delinsAAT | p.Lys184fsTer218 | 5 | 1 | |
| c.930G>C | p.Trp310Cys | 7 | 3 | |
| c.1114G>A | p.Asp372Asn | 8 | 1,2 | |
| c.1160C>G | p.Ala387Gly | 8 | 1 | |
| c.1186G>T | p.Val396Leu | 9 | 1 | |
| c.1302insA | p.Tyr434Ter | 10 | 1 | |
| c.1433C>G | p.Ala478Gly | 11 | 1 | |
| c.1658C>T | p.Thr553Ile | 13 | 1 | |
| c.1674C>G | p.Phe558Leu | 13 | 1,2 | |
| c.1741insA | p.Val581SerfsTer28 | 13 | 1 | |
| c.1795C>A | p.Gln599Lys | 14 | 1 | |
| c.1797G>C | p.Gln599His | 14 | 1 | |
| c.1804A>C | p.Ser602Arg | 14 | 1 | |
| c.1907C>A | p.Ser636Tyr | 14 | 2 | |
| c.1930G>A | p.Glu644Lys | 15 | 1 | |
| c.1935G>C | p.Leu645Phe | 15 | 1 | |
| c.2005C>G | p.Arg669Gly | 15 | 1 | |
| c.2011T>C | p.Trp671Arg | 15 | 1 | |
| c.2015G>A | p.Gly672Glu | 15 | 1,2 | |
| c.2039T>C | p.Met680Thr | 15 | 1 | |
| c.2045G>A | p.Arg682Gln | 15 | 1 | |
| c.2098A>T | p.Ile700Phe | 16 | 1 | |
| c.2171G>A | p.Cys724Tyr | 16 | 2 | |
| c.2324A>T | p.Asn775Ile | 17 | 1,2 | |

| DNA Nucleotide Change | Predicted Protein Change | Exon | Reference(s) | Reference Sequences |
|---|---|---|---|---|
| c.2350G>A | p.Gly784Ser | 17 | 1 | |
| c.2381A>C | p.Gln794Pro | 18 | 4 | |
| c.2411T>C | p.Ile804Thr | 18 | 3 | |
| c.2542T>C | p.Phe848Leu | 19 | 1 | |
| c.2547C>G | p.Phe849Leu | 19 | 1 | |
| c.2549A>G | p.His850Arg | 19 | 4 | |
| c.2554A>G | p.Met852Val | 19 | 1,2,5 | |
| c.2618G>A | p.Arg873Gln | 20 | 1 | |
| c.2660A>T | p.Asp887Val | 20 | 1 | |
| c.2690T>A | p.Ile897Asn | 20 | 6 | |
| c.2710G>A | p.Gly904Arg | 20 | 4 | |
| c.2810A>T | p.Glu937Val | 21 | 1 | |
| c.2821A>C | p.Ser941Arg | 21 | 1 | |
| c.2830G>T | p.Glu944Ter | 21 | 1,2 | |
| c.3013C>T | p.Arg1005Cys | 23 | 1,2,6 | |
| c.3014G>A | p.Arg1005His | 23 | 1,7 | |
| c.3205C>T | p.Arg1069Trp | 24 | 4 | |
| c.3407G>A | p.Arg1136Gln | 26 | 1 | |
| c.3718G>A | p.Gly1240Ser | 28 | 1 | |
| c.3742insACC | p.1248insThr | 28 | 1,2 | |
| c.3745A>C | p.Asn1249His | 28 | 5 | |
| c.3781G>A | p.Glu1261Lys | 29 | 1 | |
| c.3991G>A | p.Ala1331Thr | 30 | 1,7 | |
| c.4006C>T | p.Gln1336Ter | 30 | 2 | |
| c.-35C>G | | 5' UTR | 4 | |
| c.1048+1G>A | | Intron 7 | 4 | |
| c.1289+3A>C | | intron 9 | 4 | |
| c.1771-6C>G | | intron 13 | 4,8 | |
| c.1909+18G>A | p.Tyr637CysfsTer650 | intron15 | 2 | |
| c.1909+22G>A | | intron15 | 4 | |
| c.2617-1G>A | p.Arg873AlafsTer878 | Intron 19 | 1,2 | |
| c.2988+1G>T | | intron 21 | 1 | |

Note on variant classification: Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.

**Table 4. Selected *POLR3B* Pathogenic Variants**

| DNA Nucleotide Change | Predicted Protein Change | Exon | Reference(s) | Reference Sequences |
|---|---|---|---|---|
| c.79T>C | p.Trp27Arg | 2 | 1 | NM_018082.5 NP_060552.4 |
| c.308G>A | p.Arg103His | 6 | 1 | |
| c.312G>T | p.Leu104Phe | 6 | 1 | |
| c.802A>G | p.Ser268Gly | 10 | 1 | |
| c.832_833dup | p.Thr279SerfsTer7 | 10 | 1 | |
| c.1018C>T | p.Arg340Ter | 12 | 1 | |
| c.1112_1113delTT | p.Leu371fs | 13 | 1 | |
| c.1244T>C | p.Met415Thr | 13 | 4 | |
| c.1253C>T | p.Ala418Val | 13 | 1 | |
| c.1324C>T | p.Arg442Cys | 14 | 1 | |
| c.1325G>T | p.Arg442Leu | 14 | 1 | |
| c.1346T>C | p.Leu449Pro | 14 | 1 | |
| c.1648C>T | p.Arg550Ter | 14 | 6 | |
| c.1477G>T | p.Val493Phe | 15 | 1 | |
| c.1508C>A | p.Thr503Lys | 15 | 1,9 | |
| c.1533delT | p.Ile511MetfsTer3 | 15 | 1,9 | |
| c.1568T>A | p.Val523Glu | 15 | 1,9-11 | |
| c.1579T>C | p.Cys527Arg | 15 | 9,11 | |
| c.1788C>A | p.Tyr596Ter | 17 | 1 | |
| c.1900G>A | p.Asp634Asn | 18 | 1 | |
| c.1939G >A | p.Glu647Lys | 18 | 12 | |
| c.1999G>A | p.Val667Met | 19 | 1 | |
| c.2180T>C | p.Leu727Ser | 20 | 1 | |
| c.2190delT | p.Phe730fs | 20 | 1 | |
| c.2302C>T | p.Arg768Cys | 21 | 4 | |
| c.2303G>A | p.Arg768His | 21 | 6 | |
| c.2683G>A | p.Asp895Asn | 23 | 1 | |
| c.2686A>T | p.Lys896Ter | 23 | 1,9 | |
| c.2707delC | p.Gln903fsTer | 23 | 1 | |
| c.2774C>T | p.Pro925Leu | 24 | 4 | |
| c.2778C>G | p.Asp926Glu | 24 | 6 | |
| c.2899A>C | p.Ser967Arg | 25 | 1 | |
| c.2918G>T | p.Cys973Phe | 25 | 1 | |
| c.2920G>T | p.Glu974Ter | 25 | 1 | |
| c.2944A>G | p.Asn982Asp | 25 | 1,13 | |

| DNA Nucleotide Change | Predicted Protein Change | Exon | Reference(s) | Reference Sequences |
|---|---|---|---|---|
| c.3005T>C | p.Ile1002Thr | 26 | 1 | |
| c.3008A>G | p.Tyr1003Cys | 26 | 1,10 | |
| c.3035T>C | p.Leu1012Pro | 26 | 1 | |
| c.3071C>T | p.Ala1024Val | 26 | 1,13 | |
| c.3349C>G | p.Leu1117Val | 28 | 1 | |
| c.3352C>T | p.Gln1118Ter | 28 | 1 | |
| c.303+1G>A | | intron 5 | 1 | |
| c.967-15A>G | | intron 11 | 1 | |
| c.1101+1G>C | | intron 12 | 1 | |
| c.1263+2T>C | | intron 13 | 1 | |
| c.1464+1G>A | | intron 14 | 1 | |
| c.1857-12A>G | | intron 17 | 1 | |
| c.1857-2A>C | p.Asn620_Lys652del | intron 17 | 6 | |
| c.2083+1G>A | | intron 19 | 1 | |
| c.2084-6A>G | p.G695VfsTer5 | Intron 19 | 1,4,12 | |
| c.2570+1G>A | p.Gly818AlafsTer13 | intron 22 | 1 | |
| c.2817+30T>A | | intron 24 | 1 | |
| Del exons 21-22 | | | 10 | |
| Del exons 26-27 | | | 10 | |

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**Table 5. Selected *POLR1C* Pathogenic Variants**

| DNA Nucleotide Change | Predicted Protein Change | Exon | Reference | Reference Sequences |
|---|---|---|---|---|
| c.77C>T | p.Thr26Ile | 2 | 14 | NM_203290.3<br>NP_976035 |
| c.95A>T | p.Asn32Ile | 2 | | |
| c.193A>G | p.Met65Val | 3 | | |
| c.221A>G | p.Asn74Ser | 3 | | |
| c.281T>C | p.Val94Ala | 4 | | |
| c.326G>A | p.Arg109His | 4 | | |
| c.395G>A | p.Gly132Asp | 5 | | |
| c.436T>C | p.Cys146Arg | 5 | | |
| c.461_462delAA | p.Lys154ArgfsTer4 | 5 | | |
| c.572G>A | p.Arg191Gln | 6 | | |
| c.785T>C | p.Ile262Thr | 7 | | |
| c.883_885delAAG | p.Lys295del | 8 | | |
| c.970G4>A | p.Glu324Lys | 9 | | |

Note on variant classification: Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.

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