| Gene | Position | Mutation | Classification | Transcript | Clinical Significance |
|---|---|---|---|---|---|
| ACVRL1 | 237 | -46C>G | single nucleotide variant | NM_000020.2(ACVRL1):c.-46C>G | Benign |
| ACVRL1 | 278 | -5-33C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.-5-33C>T | Benign |
| ACVRL1 | 371 | 88C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.88C>T (p.Pro30Ser) | Likely benign |
| ACVRL1 | 490 | 207C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.207C>T (p.Cys69=) | Likely benign |
| ACVRL1 | 596 | 313+40G>C | single nucleotide variant | NM_000020.2(ACVRL1):c.313+40G>C | Benign |
| ACVRL1 | 596 | 313+11C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.313+11C>T | Benign |
| ACVRL1 | 596 | c.313+20C>A | single nucleotide variant | c.313+20C>A | Pathogenic |
| ACVRL1 | 597 | 314-35A>G | single nucleotide variant | NM_000020.2(ACVRL1):c.314-35A>G | Benign |
| ACVRL1 | 613 | 330G>A | single nucleotide variant | NM_000020.2(ACVRL1):c.330G>A (p.Ser110=) | Benign |
| ACVRL1 | 885 | c.602A > G | single nucleotide variant | c.602A > G | Pathogenic |
| ACVRL1 | 909 | 626-59delG | Deletion | NM_000020.2(ACVRL1):c.626-59delG | Likely benign |
| ACVRL1 | 925 | 642C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.642C>T (p.Gly214=) | Likely benign |
| ACVRL1 | 1030 | 747G>A | single nucleotide variant | NM_000020.2(ACVRL1):c.747G>A (p.Val249=) | Benign |
| ACVRL1 | 1055 | 772+6G>A | single nucleotide variant | NM_000020.2(ACVRL1):c.772+6G>A | Likely benign |
| ACVRL1 | 1055 | 772+27G>C | single nucleotide variant | NM_000020.2(ACVRL1):c.772+27G>C | Benign |
| ACVRL1 | 1219 | 936C>G | single nucleotide variant | 936C>G | Pathogenic |
| ACVRL1 | 1233 | c.950 T > C | single nucleotide variant | c.950 T > C | Pathogenic |
| ACVRL1 | 1405 | 1122G>T | single nucleotide variant | NM_000020.2(ACVRL1):c.1122G>T (p.Arg374=) | Benign |
| ACVRL1 | 1414 | 1131A>G | single nucleotide variant | NM_000020.2(ACVRL1):c.1131A>G (p.Ala377=) | Benign |
| ACVRL1 | 1425 | 1142T>C | single nucleotide variant | 1142T>C | Pathogenic |

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|---|---|---|---|---|---|
| ACVRL1 | 1479 | c.1196G > T | single nucleotide variant | c.1196G > T | Pathogenic |
| ACVRL1 | 1529 | 1246+9C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.1246+9C>T | Benign |
| ACVRL1 | 1529 | 1246+19C>T | single nucleotide variant | NM_000020.2(ACVRL1):c.1246+19C>T | Benign |
| ACVRL1 | 1530 | 1247-15A>G | single nucleotide variant | NM_000020.2(ACVRL1):c.1247-15A>G | Likely benign |
| ACVRL1 | 1553 | 1270C>A | single nucleotide variant | 1270C>A | Pathogenic |
| ACVRL1 | 1563 | c.1280A>T | single nucleotide variant | c.1280A>T | Pathogenic |
| ACVRL1 | 1607 | c.1324G > A | single nucleotide variant | c.1324G > A | Pathogenic |
| ACVRL1 | 1660 | 1377+45T>C | single nucleotide variant | NM_000020.2(ACVRL1):c.1377+45T>C | Likely benign |
| ACVRL1 | 1671 | c.1388 del | Deletion | c.1388 del | Pathogenic |
| ACVRL1 | 1671 | c.1388delG | Deletion | c.1388delG | Pathogenic |
| ACVRL1 | 1673 | c.1390 del | Deletion | c.1390 del | Pathogenic |
| ACVRL1 | 1719 | 1436G>A | single nucleotide variant | 1436G>A | Pathogenic |
| ACVRL1 | 1734 | c.1451 G > | single nucleotide variant | c.1451 G > | Pathogenic |
| ACVRL1 | 1734 | 1451G>A | single nucleotide variant | 1451G>A | Pathogenic |
| ACVRL1 | 1734 | 1451G>A | single nucleotide variant | 1451G>A | Pathogenic |
| ACVRL1 | 1785 | c.1502+7A>G | single nucleotide variant | c.1502+7A>G | Pathogenic |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*2422A>G | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*2398G>A | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*2380C>G | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*2313G>A | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*2281G>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1926T>C | Likely benign |

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|---|---|---|---|---|---|
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1776C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1560A>C | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1246T>C | Benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1042C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1042C>G | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1041G>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*1021T>C | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*992A>G | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*949C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*935G>C | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*913C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*869C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*862G>A | Likely benign |
| ACVRL1 | NA | | Duplication | NM_000020.2(ACVRL1):c.*856dupT | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*560T>C | Benign |

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|---|---|---|---|---|---|
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*499T>G | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*423C>T | Likely benign |
| ACVRL1 | NA | | single nucleotide variant | NM_000020.2(ACVRL1):c.*58G>A | Likely benign |
| ACVRL1 | NA | | Deletion | NM_000020.2(ACVRL1):c.*45delG | Likely benign |
| AGTR1 | 204 | -137C>A | single nucleotide variant | NM_031850.3(AGTR1):c.-137C>A | Likely benign |
| AGTR1 | 1019 | 678C>T | single nucleotide variant | NM_000685.4(AGTR1):c.573C>T (p.Leu191=) | Benign |
| AGTR1 | 1507 | c.1166 A>C | single nucleotide variant | c.1166 A>C | Pathogenic |
| AGTR1 | 1508 | 1167A>G | single nucleotide variant | NM_000685.4(AGTR1):c.1062A>G (p.Pro354=) | Benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*899T>G | Likely benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*798G>A | Benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*461C>A | Likely benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*437G>T | Likely benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*317G>A | Likely benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*135C>T | Likely benign |
| AGTR1 | NA | | single nucleotide variant | NM_031850.3(AGTR1):c.*70T>G | Likely benign |
| BMPR1B | 366 | 92G>A | single nucleotide variant | NM_001203.2(BMPR1B):c.92G>A (p.Arg31His) | Likely benign |
| BMPR1B | 521 | 247-9C>T | single nucleotide variant | NM_001203.2(BMPR1B):c.247-9C>T | Likely benign |
| BMPR1B | 521 | 247-4A>G | single nucleotide variant | NM_001203.2(BMPR1B):c.247-4A>G | Benign |
| BMPR1B | 692 | 418G>A | single nucleotide variant | NM_001203.2(BMPR1B):c.418G>A (p.Val140Ile) | Benign |
| BMPR1B | 719 | 445C>T | single nucleotide variant | NM_001203.2(BMPR1B):c.445C>T (p.Arg149Trp) | Likely benign |
| BMPR1B | 753 | c.479 G>A S160N | single nucleotide variant | c.479 G>A S160N | Pathogenic |
| BMPR1B | 753 | c.479G>A S160N | single nucleotide variant | c.479G>A S160N | Pathogenic |

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|---|---|---|---|---|---|
| BMPR1B | 945 | 671G>A | single nucleotide variant | NM_001203.2(BMPR1B):c.671G>A (p.Arg224His) | Likely benign |
| BMPR1B | 979 | 705C>T | single nucleotide variant | NM_001203.2(BMPR1B):c.705C>T (p.Thr235=) | Likely benign |
| BMPR1B | 1165 | 891C>T | single nucleotide variant | NM_001203.2(BMPR1B):c.891C>T (p.Asp297=) | Likely benign |
| BMPR1B | 1339 | 1065T>A | single nucleotide variant | NM_001203.2(BMPR1B):c.1065T>A (p.Val355=) | Likely benign |
| BMPR1B | 1376 | 1102C>T | single nucleotide variant | NM_001203.2(BMPR1B):c.1102C>T (p.Pro368Ser) | Likely benign |
| BMPR1B | 1450 | c.1176 C>A F392L | single nucleotide variant | c.1176 C>A F392L | Pathogenic |
| BMPR1B | 1450 | c.1176C>A F392L | single nucleotide variant | c.1176C>A F392L | Pathogenic |
| BMPR1B | 1658 | 1384-8T>C | single nucleotide variant | NM_001203.2(BMPR1B):c.1384-8T>C | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3679G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3646G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3618G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3328A>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3303G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3277A>T | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3255C>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3221T>C | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*3101A>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2776A>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2453A>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2433A>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2350C>T | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2332T>A | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2257C>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2221A>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2217T>C | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2216C>G | Likely benign |

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|---|---|---|---|---|---|
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*2125G>T | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1972C>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1971C>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1899A>G | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1559A>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1361G>A | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1113G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*1013C>T | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*989A>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*860T>C | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*641C>T | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*487G>T | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*413G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*196A>G | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*195C>T | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*141C>T | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*126G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*109G>A | Likely benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*49C>T | Benign |
| BMPR1B | NA | | single nucleotide variant | NM_001203.2(BMPR1B):c.*9G>C | Likely benign |
| BMPR2 | 218 | -930_-928dupGGC | Duplication | NM_001204.6(BMPR2):c.-930_-928dupGGC | Likely benign |
| BMPR2 | 221 | -927A>G | single nucleotide variant | NM_001204.6(BMPR2):c.-927A>G | Likely benign |
| BMPR2 | 227 | -921_-919dupAGC | Duplication | NM_001204.6(BMPR2):c.-921_-919dupAGC | Likely benign |
| BMPR2 | 444 | -704C>G | single nucleotide variant | NM_001204.6(BMPR2):c.-704C>G | Likely benign |
| BMPR2 | 838 | -310A>G | single nucleotide variant | NM_001204.6(BMPR2):c.-310A>G | Likely benign |
| BMPR2 | 847 | -301G>A | single nucleotide variant | NM_001204.6(BMPR2):c.-301G>A | Likely benign |
| BMPR2 | 936 | -212dupC | Duplication | NM_001204.6(BMPR2):c.-212dupC | Likely benign |

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|---|---|---|---|---|---|
| BMPR2 | 1055 | -93A>G | single nucleotide variant | NM_001204.6(BMPR2):c.-93A>G | Likely benign |
| BMPR2 | 1234 | 86A>G | single nucleotide variant | NM_001204.6(BMPR2):c.86A>G (p.Asn29Ser) | Likely benign |
| BMPR2 | 1748 | 600A>C | single nucleotide variant | NM_001204.6(BMPR2):c.600A>C (p.Leu200=) | Benign |
| BMPR2 | 1769 | 621+8T>C | single nucleotide variant | NM_001204.6(BMPR2):c.621+8T>C | Likely benign |
| BMPR2 | 1769 | 621+37C>G | single nucleotide variant | NM_001204.6(BMPR2):c.621+37C>G | Benign |
| BMPR2 | 1822 | 674G>A | single nucleotide variant | NM_001204.6(BMPR2):c.674G>A (p.Arg225His) | Likely benign |
| BMPR2 | 2001 | 853-21dupC | Duplication | NM_001204.6(BMPR2):c.853-21dupC | Likely benign |
| BMPR2 | 3472 | 2324G>A | single nucleotide variant | NM_001204.6(BMPR2):c.2324G>A (p.Ser775Asn) | Benign |
| BMPR2 | 3527 | 2379A>C | single nucleotide variant | NM_001204.6(BMPR2):c.2379A>C (p.Thr793=) | Likely benign |
| BMPR2 | 3896 | 2748T>C | single nucleotide variant | NM_001204.6(BMPR2):c.2748T>C (p.Leu916=) | Likely benign |
| BMPR2 | 3949 | 2801G>C | single nucleotide variant | NM_001204.6(BMPR2):c.2801G>C (p.Arg934Thr) | Likely benign |
| BMPR2 | 3959 | 2811G>A | single nucleotide variant | NM_001204.6(BMPR2):c.2811G>A (p.Arg937=) | Benign |
| BMPR2 | 4160 | 3012C>T | single nucleotide variant | NM_001204.6(BMPR2):c.3012C>T (p.Gly1004=) | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*7776C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*7496T>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*7306A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*7135A>G | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*6879A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*6695C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*6525G>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*6108G>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*6001T>C | Likely benign |

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|---|---|---|---|---|---|
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5998G>A | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5946C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5787T>G | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5627G>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5246C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5243T>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5152A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5044A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*5031C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*4905C>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*4364C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*3907G>C | Benign |
| BMPR2 | NA | | Deletion | NM_001204.6(BMPR2):c.*3823_*3825delCAA | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*3353T>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*3350G>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2939A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2901A>C | Likely benign |
| BMPR2 | NA | | Duplication | NM_001204.6(BMPR2):c.*2901dupA | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2833G>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2769T>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2519A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*2173A>G | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1999T>G | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1732T>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1466T>G | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1306A>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1236C>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*1226C>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*995G>A | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*815C>T | Likely benign |

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|---|---|---|---|---|---|
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*806C>T | Benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*378T>G | Likely benign |
| BMPR2 | NA | | Deletion | NM_001204.6(BMPR2):c.*309delA | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*221C>T | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*130G>C | Likely benign |
| BMPR2 | NA | | single nucleotide variant | NM_001204.6(BMPR2):c.*106C>T | Likely benign |
| CAV1 | 308 | 30+14G>T | single nucleotide variant | NM_001753.4(CAV1):c.30+14G>T | Benign |
| CAV1 | 308 | 30+19dupG | Duplication | NM_001753.4(CAV1):c.30+19dupG | Benign |
| CAV1 | 434 | 156C>T | single nucleotide variant | NM_001753.4(CAV1):c.156C>T (p.Val52=) | Benign |
| CAV1 | 741 | c.463G_A | single nucleotide variant | c.463G_A | Pathogenic |
| CAV1 | 751 | c.473delC | Deletion | c.473delC | Pathogenic |
| CAV1 | 752 | c.474delA | Deletion | c.474delA | Pathogenic |
| EDN1 | 468 | c.-134delA | Deletion | c.-134delA | Pathogenic |
| EDN1 | 928 | c.594G>T K198N | single nucleotide variant | c.594G>T K198N | Pathogenic |
| EDNRA | 1499 | c.969T>C H323H | single nucleotide variant | c.969T>C H323H | Pathogenic |
| EDNRA | 1499 | c.969T>C | single nucleotide variant | c.969T>C | Pathogenic |
| EIF2AK4 | 171 | 99T>C | single nucleotide variant | NM_001013703.3(EIF2AK4):c.99T>C (p.Ile33=) | Benign |
| EIF2AK4 | 1393 | 1321A>C | single nucleotide variant | NM_001013703.3(EIF2AK4):c.1321A>C (p.Ile441Leu) | Benign |
| EIF2AK4 | 1739 | 1667A>G | single nucleotide variant | NM_001013703.3(EIF2AK4):c.1667A>G (p.Glu556Gly) | Benign |
| EIF2AK4 | 2280 | 2208_2213dupCGACGA | Duplication | NM_001013703.3(EIF2AK4):c.2208_2213 dupCGACGA (p.Asp737_Glu738insAspAsp) | Benign |
| EIF2AK4 | 2321 | 2249+7G>A | single nucleotide variant | NM_001013703.3(EIF2AK4):c.2249+7G> A | Benign |
| EIF2AK4 | 3416 | c.3344C>T(p.P1115L) | single nucleotide variant | c.3344C>T(p.P1115L) | Pathogenic |
| EIF2AK4 | 3480 | 3408-14G>C | single nucleotide variant | NM_001013703.3(EIF2AK4):c.3408- 14G>C | Benign |

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|---|---|---|---|---|---|
| EIF2AK4 | 3988 | 3916G>T | single nucleotide variant | NM_001013703.3(EIF2AK4):c.3916G>T (p.Gly1306Cys) | Benign |
| EIF2AK4 | 4246 | 4174-17G>T | single nucleotide variant | NM_001013703.3(EIF2AK4):c.4174-17G>T | Benign |
| EIF2AK4 | 4287 | 4215C>T | single nucleotide variant | NM_001013703.3(EIF2AK4):c.4215C>T (p.Gly1405=) | Benign |
| EIF2AK4 | 4634 | 4562-8G>T | single nucleotide variant | NM_001013703.3(EIF2AK4):c.4562-8G>T | Benign |
| EIF2AK4 | NA | | single nucleotide variant | NM_001013703.3(EIF2AK4):c.*1380G>A | Benign |
| EIF2AK4 | NA | | single nucleotide variant | NM_001013703.3(EIF2AK4):c.*1290G>C | Benign |
| EIF2AK4 | NA | | single nucleotide variant | NM_001013703.3(EIF2AK4):c.*16G>A | Benign |
| EIF2AK4 | NA | | single nucleotide variant | NM_001013703.3(EIF2AK4):c.*5A>G | Benign |
| ENG | 303 | -115G>C | single nucleotide variant | NM_000118.3(ENG):c.-115G>C | Likely benign |
| ENG | 364 | -54C>A | single nucleotide variant | NM_000118.3(ENG):c.-54C>A | Likely benign |
| ENG | 409 | -9G>A | single nucleotide variant | NM_000118.3(ENG):c.-9G>A | Likely benign |
| ENG | 432 | 14C>T | single nucleotide variant | NM_000118.3(ENG):c.14C>T (p.Thr5Met) | Benign |
| ENG | 440 | c. 22T>C | single nucleotide variant | c. 22T>C | Pathogenic |
| ENG | 486 | 68-9C>G | single nucleotide variant | NM_000118.3(ENG):c.68-9C>G | Likely benign |
| ENG | 538 | 120C>T | single nucleotide variant | NM_000118.3(ENG):c.120C>T (p.Gly40=) | Benign |
| ENG | 568 | 150G>A | single nucleotide variant | NM_000118.3(ENG):c.150G>A (p.Ser50=) | Likely benign |
| ENG | 577 | 159C>T | single nucleotide variant | NM_000118.3(ENG):c.159C>T (p.Cys53=) | Benign |

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|---|---|---|---|---|---|
| ENG | 597 | 179_180delCCinsAA | Indel | NM_000118.3(ENG):c.179_180delCCinsAA (p.Ala60Glu) | Likely benign |
| ENG | 625 | 207G>A | single nucleotide variant | NM_000118.3(ENG):c.207G>A (p.Leu69=) | Benign |
| ENG | 637 | 219+25G>T | single nucleotide variant | NM_000118.3(ENG):c.219+25G>T | Benign |
| ENG | 637 | 219+22C>T | single nucleotide variant | NM_000118.3(ENG):c.219+22C>T | Benign |
| ENG | 643 | 225G>A | single nucleotide variant | NM_000118.3(ENG):c.225G>A (p.Pro75=) | Benign |
| ENG | 650 | 232C>T | single nucleotide variant | NM_000118.3(ENG):c.232C>T (p.Leu78=) | Likely benign |
| ENG | 709 | 291G>C | single nucleotide variant | NM_000118.3(ENG):c.291G>C (p.Leu97=) | Likely benign |
| ENG | 778 | 360+21C>T | single nucleotide variant | NM_000118.3(ENG):c.360+21C>T | Benign |
| ENG | 806 | 388C>T | single nucleotide variant | NM_000118.3(ENG):c.388C>T (p.Pro130Ser) | Likely benign |
| ENG | 862 | 444G>T | single nucleotide variant | NM_000118.3(ENG):c.444G>T (p.Glu148Asp) | Likely benign |
| ENG | 916 | c.498G>A | single nucleotide variant | c.498G>A | Pathogenic |
| ENG | 942 | 524-15C>T | single nucleotide variant | NM_000118.3(ENG):c.524-15C>T | Likely benign |
| ENG | 990 | 572G>A | single nucleotide variant | NM_000118.3(ENG):c.572G>A (p.Gly191Asp) | Benign |
| ENG | 1058 | c.640 G > A | single nucleotide variant | c.640 G > A | Pathogenic |
| ENG | 1192 | 774C>T | single nucleotide variant | NM_000118.3(ENG):c.774C>T (p.Tyr258=) | Likely benign |
| ENG | 1235 | 817-79C>A | single nucleotide variant | NM_000118.3(ENG):c.817-79C>A | Likely benign |
| ENG | 1327 | 909C>T | single nucleotide variant | NM_000118.3(ENG):c.909C>T (p.Ala303=) | Likely benign |

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|---|---|---|---|---|---|
| ENG | 1409 | 991+26_991+27insCCTCCC | Insertion | NM_000118.3(ENG):c.991+26_991+27insCCTCCC | Benign |
| ENG | 1417 | 999G>A | single nucleotide variant | NM_000118.3(ENG):c.999G>A (p.Arg333=) | Likely benign |
| ENG | 1432 | 1014C>T | single nucleotide variant | NM_000118.3(ENG):c.1014C>T (p.Pro338=) | Likely benign |
| ENG | 1438 | 1020G>A | single nucleotide variant | NM_000118.3(ENG):c.1020G>A (p.Pro340=) | Likely benign |
| ENG | 1447 | 1029C>T | single nucleotide variant | NM_000118.3(ENG):c.1029C>T (p.Thr343=) | Benign |
| ENG | 1478 | 1060C>T | single nucleotide variant | NM_000118.3(ENG):c.1060C>T (p.Leu354=) | Benign |
| ENG | 1513 | 1095C>T | single nucleotide variant | NM_000118.3(ENG):c.1095C>T (p.Asp365=) | Likely benign |
| ENG | 1514 | 1096G>C | single nucleotide variant | NM_000118.3(ENG):c.1096G>C (p.Asp366His) | Benign |
| ENG | 1516 | 1098C>T | single nucleotide variant | NM_000118.3(ENG):c.1098C>T (p.Asp366=) | Likely benign |
| ENG | 1691 | 1273-4G>A | single nucleotide variant | NM_000118.3(ENG):c.1273-4G>A | Likely benign |
| ENG | 1792 | 1374A>G | single nucleotide variant | NM_000118.3(ENG):c.1374A>G (p.Pro458=) | Benign |
| ENG | 1864 | 1446C>T | single nucleotide variant | NM_000118.3(ENG):c.1446C>T (p.Ser482=) | Likely benign |
| ENG | 1865 | 1447G>A | single nucleotide variant | NM_000118.3(ENG):c.1447G>A (p.Val483Ile) | Likely benign |
| ENG | 1870 | 1452C>T | single nucleotide variant | NM_000118.3(ENG):c.1452C>T (p.Ser484=) | Likely benign |
| ENG | 1873 | 1455G>A | single nucleotide variant | NM_000118.3(ENG):c.1455G>A (p.Glu485=) | Likely benign |

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|---|---|---|---|---|---|
| ENG | 1951 | 1533G>A | single nucleotide variant | NM_000118.3(ENG):c.1533G>A (p.Ala511=) | Benign |
| ENG | 2051 | c.1633 G > A | single nucleotide variant | c.1633 G > A | Pathogenic |
| ENG | 2105 | 1687-17C>T | single nucleotide variant | NM_000118.3(ENG):c.1687-17C>T | Likely benign |
| ENG | 2105 | 1687-7C>T | single nucleotide variant | NM_000118.3(ENG):c.1687-7C>T | Likely benign |
| ENG | 2130 | 1712G>A | single nucleotide variant | NM_000118.3(ENG):c.1712G>A (p.Arg571His) | Likely benign |
| ENG | 2212 | 1794T>C | single nucleotide variant | NM_000118.3(ENG):c.1794T>C (p.Gly598=) | Benign |
| ENG | 2262 | 1844C>T | single nucleotide variant | NM_000118.3(ENG):c.1844C>T (p.Ser615Leu) | Likely benign |
| ENG | 2263 | 1845G>T | single nucleotide variant | NM_000118.3(ENG):c.1845G>T (p.Ser615=) | Likely benign |
| ENG | NA | | Deletion | NM_000118.3(ENG):c.*700_*703delAACT | Likely benign |
| ENG | NA | | single nucleotide variant | NM_001114753.2(ENG):c.1932C>T (p.Ile644=) | Benign |
| KCNA5 | 62 | -167C>T | single nucleotide variant | NM_002234.3(KCNA5):c.-167C>T | Likely benign |
| KCNA5 | 321 | 92G>T | single nucleotide variant | NM_002234.3(KCNA5):c.92G>T (p.Gly31Val) | Likely benign |
| KCNA5 | 354 | c.125T>A | single nucleotide variant | c.125T>A | Pathogenic |
| KCNA5 | 411 | 182 G>A (G182R) | single nucleotide variant | 182 G>A (G182R) | Pathogenic |
| KCNA5 | 440 | 211 G>C (E211D) | single nucleotide variant | 211 G>C (E211D) | Pathogenic |
| KCNA5 | 482 | c.253C>A | single nucleotide variant | c.253C>A | Pathogenic |
| KCNA5 | 569 | c.340A>C | single nucleotide variant | c.340A>C | Pathogenic |
| KCNA5 | 594 | c.-365C>T | single nucleotide variant | c.-365C>T | Pathogenic |
| KCNA5 | 610 | 381C>T | single nucleotide variant | NM_002234.3(KCNA5):c.381C>T (p.Ser127=) | Likely benign |
| KCNA5 | 614 | c.385C>G | single nucleotide variant | c.385C>G | Pathogenic |
| KCNA5 | 738 | c.509C>G | single nucleotide variant | c.509C>G | Pathogenic |
| KCNA5 | 773 | c.544G>A (p.G182R) | single nucleotide variant | c.544G>A (p.G182R) | Pathogenic |

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|---|---|---|---|---|---|
| KCNA5 | 780 | c.551G>C(p.(R184P)) | single nucleotide variant | c.551G>C(p.(R184P)) | Pathogenic |
| KCNA5 | 862 | c.633G>C(p.Glu211Asp) | single nucleotide variant | c.633G>C(p.Glu211Asp) | Pathogenic |
| KCNA5 | 980 | 751G>A | single nucleotide variant | NM_002234.3(KCNA5):c.751G>A (p.Ala251Thr) | Benign |
| KCNA5 | 1158 | 929C>T | single nucleotide variant | NM_002234.3(KCNA5):c.929C>T (p.Pro310Leu) | Benign |
| KCNA5 | 1378 | 1149T>C | single nucleotide variant | NM_002234.3(KCNA5):c.1149T>C (p.Gly383=) | Benign |
| KCNA5 | 1896 | p.Y483* | Deletion | p.Y483* | Pathogenic |
| KCNA5 | 1962 | 1733G>A | single nucleotide variant | NM_002234.3(KCNA5):c.1733G>A (p.Arg578Lys) | Benign |
| KCNA5 | 1962 | c.1733G>A | single nucleotide variant | c.1733G>A | Pathogenic |
| KCNA5 | 2071 | c.1842+508A>T | single nucleotide variant | c.1842+508A>T | Pathogenic |
| KCNA5 | NA | | single nucleotide variant | NM_002234.3(KCNA5):c.*800G>A | Likely benign |
| KCNA5 | NA | | single nucleotide variant | NM_002234.3(KCNA5):c.*736A>T | Likely benign |
| KCNA5 | NA | | single nucleotide variant | NM_002234.3(KCNA5):c.*508A>T | Likely benign |
| KCNK3 | 249 | p.T29K | single nucleotide variant | p.T29K | Pathogenic |
| KCNK3 | 515 | p.G118R | single nucleotide variant | p.G118R | Pathogenic |
| KCNK3 | 738 | p.Y192C | single nucleotide variant | p.Y192C | Pathogenic |
| KCNK3 | 770 | p.G203D | single nucleotide variant | p.G203D | Pathogenic |
| KCNK3 | 771 | c.608 G>A (G203D) | single nucleotide variant | c.608 G>A (G203D) | Pathogenic |
| KCNK3 | 824 | p.V221L | single nucleotide variant | p.V221L | Pathogenic |
| KCNK3 | NA | | single nucleotide variant | NM_002246.2(KCNK3):c.*13G>A | Benign |
| NOS2 | 664 | c.-400delCCTTT | single nucleotide variant | c.-400delCCTTT | Pathogenic |
| NOTCH3 | 196 | 120C>G | single nucleotide variant | NM_000435.2(NOTCH3):c.120C>G (p.Ala40=) | Likely benign |
| NOTCH3 | 379 | 303C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.303C>T (p.Thr101=) | Benign |

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|---|---|---|---|---|---|
| NOTCH3 | 414 | 338G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.338G>A (p.Arg113Gln) | Likely benign |
| NOTCH3 | 585 | 509A>G | single nucleotide variant | NM_000435.2(NOTCH3):c.509A>G (p.His170Arg) | Likely benign |
| NOTCH3 | 682 | 606A>G | single nucleotide variant | NM_000435.2(NOTCH3):c.606A>G (p.Ala202=) | Benign |
| NOTCH3 | 736 | 660C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.660C>T (p.Tyr220=) | Likely benign |
| NOTCH3 | 1021 | 945C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.945C>T (p.Ile315=) | Likely benign |
| NOTCH3 | 1040 | 964G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.964G>A (p.Val322Met) | Likely benign |
| NOTCH3 | 1216 | 1140T>C | single nucleotide variant | NM_000435.2(NOTCH3):c.1140T>C (p.Pro380=) | Benign |
| NOTCH3 | 1268 | 1192+15A>G | single nucleotide variant | NM_000435.2(NOTCH3):c.1192+15A>G | Benign |
| NOTCH3 | 1563 | 1487C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.1487C>T (p.Pro496Leu) | Benign |
| NOTCH3 | 1566 | 1490C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.1490C>T (p.Ser497Leu) | Benign |

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|---|---|---|---|---|---|
| NOTCH3 | 1696 | 1620G>T | single nucleotide variant | NM_000435.2(NOTCH3):c.1620G>T (p.Thr540=) | Likely benign |
| NOTCH3 | 1766 | 1690G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.1690G>A (p.Ala564Thr) | Likely benign |
| NOTCH3 | 1801 | 1725G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.1725G>A (p.Thr575=) | Benign |
| NOTCH3 | 1858 | 1782C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.1782C>T (p.Gly594=) | Benign |
| NOTCH3 | 1916 | 1840+19G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.1840+19G>A | Likely benign |
| NOTCH3 | 2115 | 2039G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.2039G>A (p.Arg680His) | Benign |
| NOTCH3 | 2278 | 2202C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.2202C>T (p.Ala734=) | Benign |
| NOTCH3 | 2376 | 2300G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.2300G>A (p.Arg767His) | Benign |
| NOTCH3 | 2487 | 2411-4C>G | single nucleotide variant | NM_000435.2(NOTCH3):c.2411-4C>G | Likely benign |
| NOTCH3 | 2595 | c.2519G>A p.G840E | single nucleotide variant | c.2519G>A p.G840E | Pathogenic |
| NOTCH3 | 2614 | 2538C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.2538C>T (p.Cys846=) | Benign |
| NOTCH3 | 2656 | 2580C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.2580C>T (p.Asn860=) | Likely benign |

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|---|---|---|---|---|---|
| NOTCH3 | 2774 | c.2698A>C p.T900P | single nucleotide variant | c.2698A>C p.T900P | Pathogenic |
| NOTCH3 | 2818 | 2742A>G | single nucleotide variant | NM_000435.2(NOTCH3):c.2742A>G (p.Pro914=) | Benign |
| NOTCH3 | 3475 | 3399C>A | single nucleotide variant | NM_000435.2(NOTCH3):c.3399C>A (p.His1133Gln) | Benign |
| NOTCH3 | 3537 | 3461-15C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.3461-15C>T | Likely benign |
| NOTCH3 | 3599 | 3523C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.3523C>T (p.Arg1175Trp) | Likely benign |
| NOTCH3 | 3623 | 3547G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.3547G>A (p.Val1183Met) | Benign |
| NOTCH3 | 3715 | 3639C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.3639C>T (p.Ala1213=) | Likely benign |
| NOTCH3 | 3780 | 3704A>T | single nucleotide variant | NM_000435.2(NOTCH3):c.3704A>T (p.His1235Leu) | Likely benign |
| NOTCH3 | 3795 | 3719-11C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.3719-11C>T | Likely benign |
| NOTCH3 | 4147 | 4071C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.4071C>T (p.Pro1357=) | Likely benign |
| NOTCH3 | 4267 | 4191G>T | single nucleotide variant | NM_000435.2(NOTCH3):c.4191G>T (p.Gly1397=) | Likely benign |

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|---|---|---|---|---|---|
| NOTCH3 | 4424 | 4348G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.4348G>A (p.Ala1450Thr) | Likely benign |
| NOTCH3 | 4520 | 4444G>C | single nucleotide variant | NM_000435.2(NOTCH3):c.4444G>C (p.Gly1482Arg) | Likely benign |
| NOTCH3 | 4628 | 4552C>A | single nucleotide variant | NM_000435.2(NOTCH3):c.4552C>A (p.Leu1518Met) | Likely benign |
| NOTCH3 | 4639 | 4563A>G | single nucleotide variant | NM_000435.2(NOTCH3):c.4563A>G (p.Pro1521=) | Benign |
| NOTCH3 | 4755 | 4679G>C | single nucleotide variant | NM_000435.2(NOTCH3):c.4679G>C (p.Arg1560Pro) | Benign |
| NOTCH3 | 5438 | 5362+3T>C | single nucleotide variant | NM_000435.2(NOTCH3):c.5362+3T>C | Benign |
| NOTCH3 | 5439 | 5363-17C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.5363-17C>T | Benign |
| NOTCH3 | 5446 | 5370C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.5370C>T (p.Phe1790=) | Benign |
| NOTCH3 | 5452 | 5376G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.5376G>A (p.Pro1792=) | Likely benign |
| NOTCH3 | 5476 | 5400G>T | single nucleotide variant | NM_000435.2(NOTCH3):c.5400G>T (p.Gly1800=) | Benign |
| NOTCH3 | 5542 | 5466C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.5466C>T (p.Ser1822=) | Likely benign |

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|---|---|---|---|---|---|
| NOTCH3 | 5577 | 5501G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.5501G>A (p.Arg1834Gln) | Likely benign |
| NOTCH3 | 5602 | 5526T>C | single nucleotide variant | NM_000435.2(NOTCH3):c.5526T>C (p.Ala1842=) | Benign |
| NOTCH3 | 5744 | 5668-11C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.5668-11C>T | Likely benign |
| NOTCH3 | 5892 | 5816-8T>C | single nucleotide variant | NM_000435.2(NOTCH3):c.5816-8T>C | Benign |
| NOTCH3 | 5930 | 5854G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.5854G>A (p.Val1952Met) | Benign |
| NOTCH3 | 6107 | 6031G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.6031G>A (p.Val2011Ile) | Benign |
| NOTCH3 | 6178 | 6102C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6102C>T (p.Pro2034=) | Benign |
| NOTCH3 | 6242 | 6166G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.6166G>A (p.Ala2056Thr) | Likely benign |
| NOTCH3 | 6297 | 6221C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6221C>T (p.Pro2074Leu) | Benign |
| NOTCH3 | 6514 | 6438G>A | single nucleotide variant | NM_000435.2(NOTCH3):c.6438G>A (p.Ala2146=) | Benign |

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|---|---|---|---|---|---|
| NOTCH3 | 6608 | 6532C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6532C>T (p.Pro2178Ser) | Likely benign |
| NOTCH3 | 6687 | 6611C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6611C>T (p.Pro2204Leu) | Likely benign |
| NOTCH3 | 6744 | 6668C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6668C>T (p.Ala2223Val) | Benign |
| NOTCH3 | 6829 | 6753C>T | single nucleotide variant | NM_000435.2(NOTCH3):c.6753C>T (p.Ser2251=) | Benign |
| NOTCH3 | 6889 | 6813T>C | single nucleotide variant | NM_000435.2(NOTCH3):c.6813T>C (p.Pro2271=) | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*23T>A | Likely benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*96C>T | Likely benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*338A>C | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*370A>G | Likely benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*544C>T | Likely benign |

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|---|---|---|---|---|---|
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*563T>C | Likely benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*608C>T | Likely benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*668G>T | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*808G>A | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*837G>A | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*890C>T | Benign |
| NOTCH3 | NA | | single nucleotide variant | NM_000435.2(NOTCH3):c.*937T>C | Likely benign |
| SERPINE1 | 206 | 49G>A | single nucleotide variant | NM_000602.4(SERPINE1):c.49G>A (p.Val17Ile) | Likely benign |
| SERPINE1 | 712 | 555G>C | single nucleotide variant | NM_000602.4(SERPINE1):c.555G>C (p.Arg185=) | Likely benign |
| SERPINE1 | 832 | c.-675del | Deletion | c.-675del | Pathogenic |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*1737G>A | Benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*1358T>C | Likely benign |

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|---|---|---|---|---|---|
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*1186C>T | Likely benign |
| SERPINE1 | NA | | Duplication | NM_000602.4(SERPINE1):c.*997_*1005dupCCGCGCCCC | Benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*892G>A | Likely benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*752C>A | Likely benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*722T>G | Benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*690T>C | Likely benign |
| SERPINE1 | NA | | single nucleotide variant | NM_000602.4(SERPINE1):c.*361T>C | Benign |
| SIRT3 | 656 | c.622G>A | single nucleotide variant | c.622G>A | Pathogenic |
| SMAD4 | 403 | -135G>A | single nucleotide variant | NM_005359.5(SMAD4):c.-135G>A | Likely benign |
| SMAD4 | 405 | -133G>A | single nucleotide variant | NM_005359.5(SMAD4):c.-133G>A | Likely benign |
| SMAD4 | 410 | -128+16_-128+18delCCCinsTT | Indel | NM_005359.5(SMAD4):c.-128+16_-128+18delCCCinsTT | Likely benign |
| SMAD4 | 410 | -128+1995A>G | single nucleotide variant | NM_005359.5(SMAD4):c.-128+1995A>G | Likely benign |
| SMAD4 | 410 | -128+759G>A | single nucleotide variant | NM_005359.5(SMAD4):c.-128+759G>A | Likely benign |
| SMAD4 | 410 | -128+540A>G | single nucleotide variant | NM_005359.5(SMAD4):c.-128+540A>G | Likely benign |
| SMAD4 | 410 | -128+263G>C | single nucleotide variant | NM_005359.5(SMAD4):c.-128+263G>C | Likely benign |
| SMAD4 | 410 | -128+3139C>T | single nucleotide variant | NM_005359.5(SMAD4):c.-128+3139C>T | Likely benign |
| SMAD4 | 411 | -127-3T>A | single nucleotide variant | NM_005359.5(SMAD4):c.-127-3T>A | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 411 | -127-?_*6575+?dup | Duplication | NM_005359.5(SMAD4):c.-127-?_*6575+?dup | Likely benign |
| SMAD4 | 411 | -127-4838G>A | single nucleotide variant | NM_005359.5(SMAD4):c.-127-4838G>A | Likely benign |
| SMAD4 | 411 | -127-4835G>A | single nucleotide variant | NM_005359.5(SMAD4):c.-127-4835G>A | Likely benign |
| SMAD4 | 411 | -127-3318A>G | single nucleotide variant | NM_005359.5(SMAD4):c.-127-3318A>G | Likely benign |
| SMAD4 | 411 | -127-135A>C | single nucleotide variant | NM_005359.5(SMAD4):c.-127-135A>C | Likely benign |
| SMAD4 | 485 | -53_-48delAATTGC | Deletion | NM_005359.5(SMAD4):c.-53_-48delAATTGC | Likely benign |
| SMAD4 | 499 | -39T>A | single nucleotide variant | NM_005359.5(SMAD4):c.-39T>A | Likely benign |
| SMAD4 | 510 | -28T>C | single nucleotide variant | NM_005359.5(SMAD4):c.-28T>C | Likely benign |
| SMAD4 | 518 | -20A>C | single nucleotide variant | NM_005359.5(SMAD4):c.-20A>C | Likely benign |
| SMAD4 | 527 | -11A>C | single nucleotide variant | NM_005359.5(SMAD4):c.-11A>C | Likely benign |
| SMAD4 | 559 | 21G>A | single nucleotide variant | NM_005359.5(SMAD4):c.21G>A (p.Thr7=) | Likely benign |
| SMAD4 | 576 | c.38A>G, p.N13S | single nucleotide variant | c.38A>G, p.N13S | Pathogenic |
| SMAD4 | 598 | 60G>A | single nucleotide variant | NM_005359.5(SMAD4):c.60G>A (p.Val20=) | Likely benign |
| SMAD4 | 628 | 90A>G | single nucleotide variant | NM_005359.5(SMAD4):c.90A>G (p.Gly30=) | Likely benign |
| SMAD4 | 640 | 102A>G | single nucleotide variant | NM_005359.5(SMAD4):c.102A>G (p.Thr34=) | Likely benign |
| SMAD4 | 664 | 126T>C | single nucleotide variant | NM_005359.5(SMAD4):c.126T>C (p.Ser42=) | Likely benign |
| SMAD4 | 665 | c.127A>G, p.K43E | single nucleotide variant | c.127A>G, p.K43E | Pathogenic |
| SMAD4 | 670 | 132A>G | single nucleotide variant | NM_005359.5(SMAD4):c.132A>G (p.Val44=) | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 682 | 144G>A | single nucleotide variant | NM_005359.5(SMAD4):c.144G>A (p.Lys48=) | Likely benign |
| SMAD4 | 715 | 177A>G | single nucleotide variant | NM_005359.5(SMAD4):c.177A>G (p.Thr59=) | Likely benign |
| SMAD4 | 721 | 183A>C | single nucleotide variant | NM_005359.5(SMAD4):c.183A>C (p.Ile61=) | Likely benign |
| SMAD4 | 766 | 228A>G | single nucleotide variant | NM_005359.5(SMAD4):c.228A>G (p.Arg76=) | Likely benign |
| SMAD4 | 769 | 231A>G | single nucleotide variant | NM_005359.5(SMAD4):c.231A>G (p.Thr77=) | Likely benign |
| SMAD4 | 787 | 249+9T>C | single nucleotide variant | NM_005359.5(SMAD4):c.249+9T>C | Likely benign |
| SMAD4 | 788 | 250-15C>T | single nucleotide variant | NM_005359.5(SMAD4):c.250-15C>T | Likely benign |
| SMAD4 | 788 | 250-589A>G | single nucleotide variant | NM_005359.5(SMAD4):c.250-589A>G | Likely benign |
| SMAD4 | 799 | 261G>A | single nucleotide variant | NM_005359.5(SMAD4):c.261G>A (p.Arg87=) | Likely benign |
| SMAD4 | 814 | 276T>C | single nucleotide variant | NM_005359.5(SMAD4):c.276T>C (p.His92=) | Likely benign |
| SMAD4 | 880 | 342T>C | single nucleotide variant | NM_005359.5(SMAD4):c.342T>C (p.Tyr114=) | Likely benign |
| SMAD4 | 892 | 354G>A | single nucleotide variant | NM_005359.5(SMAD4):c.354G>A (p.Ala118=) | Benign |
| SMAD4 | 904 | 366A>G | single nucleotide variant | NM_005359.5(SMAD4):c.366A>G (p.Lys122=) | Likely benign |
| SMAD4 | 907 | 369T>C | single nucleotide variant | NM_005359.5(SMAD4):c.369T>C (p.Cys123=) | Likely benign |
| SMAD4 | 913 | 375T>C | single nucleotide variant | NM_005359.5(SMAD4):c.375T>C (p.Ser125=) | Likely benign |
| SMAD4 | 925 | 387T>C | single nucleotide variant | NM_005359.5(SMAD4):c.387T>C (p.Asn129=) | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 937 | 399C>T | single nucleotide variant | NM_005359.5(SMAD4):c.399C>T (p.Tyr133=) | Likely benign |
| SMAD4 | 949 | 411A>G | single nucleotide variant | NM_005359.5(SMAD4):c.411A>G (p.Val137=) | Likely benign |
| SMAD4 | 962 | 424+12_424+13delTT | Deletion | NM_005359.5(SMAD4):c.424+12_424+13delTT | Likely benign |
| SMAD4 | 962 | 424+14G>A | single nucleotide variant | NM_005359.5(SMAD4):c.424+14G>A | Likely benign |
| SMAD4 | 962 | 424+165A>G | single nucleotide variant | NM_005359.5(SMAD4):c.424+165A>G | Likely benign |
| SMAD4 | 992 | 454+291T>A | single nucleotide variant | NM_005359.5(SMAD4):c.454+291T>A | Likely benign |
| SMAD4 | 992 | 454+8A>T | single nucleotide variant | NM_005359.5(SMAD4):c.454+8A>T | Likely benign |
| SMAD4 | 993 | 455-8C>T | single nucleotide variant | NM_005359.5(SMAD4):c.455-8C>T | Likely benign |
| SMAD4 | 993 | 455-43A>T | single nucleotide variant | NM_005359.5(SMAD4):c.455-43A>T | Benign |
| SMAD4 | 993 | 455-1921T>G | single nucleotide variant | NM_005359.5(SMAD4):c.455-1921T>G | Likely benign |
| SMAD4 | 993 | 455-6A>G | single nucleotide variant | NM_005359.5(SMAD4):c.455-6A>G | Benign |
| SMAD4 | 1012 | 474G>A | single nucleotide variant | NM_005359.5(SMAD4):c.474G>A (p.Val158=) | Likely benign |
| SMAD4 | 1021 | 483A>G | single nucleotide variant | NM_005359.5(SMAD4):c.483A>G (p.Glu161=) | Likely benign |
| SMAD4 | 1024 | 486T>C | single nucleotide variant | NM_005359.5(SMAD4):c.486T>C (p.Tyr162=) | Likely benign |
| SMAD4 | 1045 | 507G>A | single nucleotide variant | NM_005359.5(SMAD4):c.507G>A (p.Gln169=) | Likely benign |
| SMAD4 | 1048 | 510A>G | single nucleotide variant | NM_005359.5(SMAD4):c.510A>G (p.Pro170=) | Likely benign |
| SMAD4 | 1120 | 582A>G | single nucleotide variant | NM_005359.5(SMAD4):c.582A>G (p.Thr194=) | Likely benign |
| SMAD4 | 1144 | 606C>G | single nucleotide variant | NM_005359.5(SMAD4):c.606C>G (p.Ala202=) | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 1171 | 633T>C | single nucleotide variant | NM_005359.5(SMAD4):c.633T>C (p.Thr211=) | Likely benign |
| SMAD4 | 1177 | 639C>T | single nucleotide variant | NM_005359.5(SMAD4):c.639C>T (p.Asn213=) | Likely benign |
| SMAD4 | 1205 | 667+20G>T | single nucleotide variant | NM_005359.5(SMAD4):c.667+20G>T | Likely benign |
| SMAD4 | 1205 | 667+17G>A | single nucleotide variant | NM_005359.5(SMAD4):c.667+17G>A | Likely benign |
| SMAD4 | 1205 | 667+9T>C | single nucleotide variant | NM_005359.5(SMAD4):c.667+9T>C | Likely benign |
| SMAD4 | 1205 | 667+54T>C | single nucleotide variant | NM_005359.5(SMAD4):c.667+54T>C | Likely benign |
| SMAD4 | 1205 | 667+299T>C | single nucleotide variant | NM_005359.5(SMAD4):c.667+299T>C | Likely benign |
| SMAD4 | 1206 | 668-7C>T | single nucleotide variant | NM_005359.5(SMAD4):c.668-7C>T | Likely benign |
| SMAD4 | 1206 | 668-1111A>G | single nucleotide variant | NM_005359.5(SMAD4):c.668-1111A>G | Likely benign |
| SMAD4 | 1231 | 693C>T | single nucleotide variant | NM_005359.5(SMAD4):c.693C>T (p.Gly231=) | Likely benign |
| SMAD4 | 1325 | 787+15T>G | single nucleotide variant | NM_005359.5(SMAD4):c.787+15T>G | Likely benign |
| SMAD4 | 1325 | 787+15T>C | single nucleotide variant | NM_005359.5(SMAD4):c.787+15T>C | Likely benign |
| SMAD4 | 1326 | 788-12delC | Deletion | NM_005359.5(SMAD4):c.788-12delC | Likely benign |
| SMAD4 | 1336 | 798C>T | single nucleotide variant | NM_005359.5(SMAD4):c.798C>T (p.Thr266=) | Likely benign |
| SMAD4 | 1363 | 825A>G | single nucleotide variant | NM_005359.5(SMAD4):c.825A>G (p.Pro275=) | Likely benign |
| SMAD4 | 1408 | 870C>T | single nucleotide variant | NM_005359.5(SMAD4):c.870C>T (p.His290=) | Likely benign |
| SMAD4 | 1414 | 876G>A | single nucleotide variant | NM_005359.5(SMAD4):c.876G>A (p.Pro292=) | Likely benign |
| SMAD4 | 1442 | 904+298T>C | single nucleotide variant | NM_005359.5(SMAD4):c.904+298T>C | Likely benign |
| SMAD4 | 1443 | 905-19_905-17delTTT | Deletion | NM_005359.5(SMAD4):c.905-19_905-17delTTT | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 1443 | 905-52A>G | single nucleotide variant | NM_005359.5(SMAD4):c.905-52A>G | Benign |
| SMAD4 | 1443 | 905-32T= | single nucleotide variant | NM_005359.5(SMAD4):c.905-32T= | Benign |
| SMAD4 | 1447 | 909T>C | single nucleotide variant | NM_005359.5(SMAD4):c.909T>C (p.Pro303=) | Likely benign |
| SMAD4 | 1459 | 921G>A | single nucleotide variant | NM_005359.5(SMAD4):c.921G>A (p.Glu307=) | Likely benign |
| SMAD4 | 1462 | 924T>C | single nucleotide variant | NM_005359.5(SMAD4):c.924T>C (p.Leu308=) | Likely benign |
| SMAD4 | 1493 | 955+15A>G | single nucleotide variant | NM_005359.5(SMAD4):c.955+15A>G | Likely benign |
| SMAD4 | 1493 | 955+7G>A | single nucleotide variant | NM_005359.5(SMAD4):c.955+7G>A | Likely benign |
| SMAD4 | 1494 | 956-5T>C | single nucleotide variant | NM_005359.5(SMAD4):c.956-5T>C | Likely benign |
| SMAD4 | 1501 | 963G>A | single nucleotide variant | NM_005359.5(SMAD4):c.963G>A (p.Glu321=) | Likely benign |
| SMAD4 | 1543 | 1005A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1005A>G (p.Val335=) | Likely benign |
| SMAD4 | 1552 | 1014A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1014A>G (p.Thr338=) | Likely benign |
| SMAD4 | 1597 | 1059C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1059C>T (p.Tyr353=) | Likely benign |
| SMAD4 | 1624 | 1086T>C | single nucleotide variant | NM_005359.5(SMAD4):c.1086T>C (p.Phe362=) | Benign |
| SMAD4 | 1663 | 1125C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1125C>T (p.Ala375=) | Likely benign |
| SMAD4 | 1678 | 1140-36_1140-19del18 | Deletion | NM_005359.5(SMAD4):c.1140-36_1140-19del18 | Likely benign |
| SMAD4 | 1678 | 1140-10delT | Deletion | NM_005359.5(SMAD4):c.1140-10delT | Likely benign |
| SMAD4 | 1678 | 1140G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1140G>A (p.Arg380=) | Likely benign |
| SMAD4 | 1678 | 1140-16C>A | single nucleotide variant | NM_005359.5(SMAD4):c.1140-16C>A | Likely benign |
| SMAD4 | 1678 | 1140-4T>C | single nucleotide variant | NM_005359.5(SMAD4):c.1140-4T>C | Likely benign |
| SMAD4 | 1678 | 1140-10T>C | single nucleotide variant | NM_005359.5(SMAD4):c.1140-10T>C | Benign |

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|---|---|---|---|---|---|
| SMAD4 | 1744 | 1206T>A | single nucleotide variant | NM_005359.5(SMAD4):c.1206T>A (p.Leu402=) | Likely benign |
| SMAD4 | 1753 | 1215C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1215C>T (p.His405=) | Likely benign |
| SMAD4 | 1756 | 1218G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1218G>A (p.Ala406=) | Likely benign |
| SMAD4 | 1774 | 1236C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1236C>T (p.Tyr412=) | Likely benign |
| SMAD4 | 1786 | 1248A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1248A>G (p.Arg416=) | Likely benign |
| SMAD4 | 1846 | 1308+10A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1308+10A>G | Likely benign |
| SMAD4 | 1846 | 1308+9C>A | single nucleotide variant | NM_005359.5(SMAD4):c.1308+9C>A | Likely benign |
| SMAD4 | 1846 | 1308+2398T>G | single nucleotide variant | NM_005359.5(SMAD4):c.1308+2398T>G | Likely benign |
| SMAD4 | 1846 | 1308+4116C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1308+4116C>T | Likely benign |
| SMAD4 | 1846 | 1308+623C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1308+623C>T | Likely benign |
| SMAD4 | 1849 | 1311C>G | single nucleotide variant | NM_005359.5(SMAD4):c.1311C>G (p.Val437=) | Likely benign |
| SMAD4 | 1909 | 1371A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1371A>G (p.Ala457=) | Likely benign |
| SMAD4 | 1930 | 1392C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1392C>T (p.Ala464=) | Likely benign |
| SMAD4 | 1933 | 1395G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1395G>A (p.Val465=) | Likely benign |
| SMAD4 | 1960 | 1422A>C | single nucleotide variant | NM_005359.5(SMAD4):c.1422A>C (p.Ser474=) | Likely benign |
| SMAD4 | 1985 | 1447+9G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1447+9G>A | Likely benign |
| SMAD4 | 1986 | 1448-113T>C | single nucleotide variant | NM_005359.5(SMAD4):c.1448-113T>C | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | 1986 | c.1448-6T>C | single nucleotide variant | c.1448-6T>C | Pathogenic |
| SMAD4 | 1999 | 1461T>A | single nucleotide variant | NM_005359.5(SMAD4):c.1461T>A (p.Ala487=) | Likely benign |
| SMAD4 | 2030 | 1492T>C | single nucleotide variant | NM_005359.5(SMAD4):c.1492T>C (p.Leu498=) | Likely benign |
| SMAD4 | 2071 | 1533G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1533G>A (p.Pro511=) | Likely benign |
| SMAD4 | 2092 | 1554C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1554C>T (p.Ile518=) | Likely benign |
| SMAD4 | 2134 | 1596C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1596C>T (p.Ala532=) | Likely benign |
| SMAD4 | 2144 | 1606C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1606C>T (p.Leu536=) | Likely benign |
| SMAD4 | 2149 | 1611C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1611C>T (p.Asp537=) | Likely benign |
| SMAD4 | 2170 | 1632G>A | single nucleotide variant | NM_005359.5(SMAD4):c.1632G>A (p.Pro544=) | Likely benign |
| SMAD4 | 2179 | 1641C>T | single nucleotide variant | NM_005359.5(SMAD4):c.1641C>T (p.Asp547=) | Likely benign |
| SMAD4 | 2185 | 1647A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1647A>G (p.Gln549=) | Likely benign |
| SMAD4 | 2191 | 1653A>G | single nucleotide variant | NM_005359.5(SMAD4):c.1653A>G (p.Leu551=) | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*12G>A | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6588C>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6586C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6513C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6492A>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6423G>C | Benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*6353delT | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*6009G>C | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5994A>C | Benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*5863_*5867delGAAAA | Benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5801T>C | Likely benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*5691_*5693delTAT | Benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*5637_*5640delACAC | Benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5627G>A | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5535A>G | Benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5419T>C | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5259A>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5170C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5131A>G | Benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5083G>A | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*5080A>G | Likely benign |
| SMAD4 | NA | | Duplication | NM_005359.5(SMAD4):c.*4867dupT | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*4862A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*4748C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*4643T>C | Likely benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*4587_*4590delGAGA | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*4378T>G | Likely benign |
| SMAD4 | NA | | Duplication | NM_005359.5(SMAD4):c.*3878dupT | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*3763C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*3398A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2989A>G | Likely benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*2968delT | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2914C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2796G>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2488T>A | Likely benign |

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|---|---|---|---|---|---|
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2361A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2353C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*2122A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1866A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1864C>A | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1820T>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1812T>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1187A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1179T>C | Likely benign |
| SMAD4 | NA | | Deletion | NM_005359.5(SMAD4):c.*1169_*1173delCCATC | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*1067G>A | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*685A>C | Benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*412A>G | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*334C>T | Likely benign |
| SMAD4 | NA | | single nucleotide variant | NM_005359.5(SMAD4):c.*11C>T | Benign |
| SMAD4 | NA | | Duplication | NM_005359.5(SMAD4):c.(?_-1)_249+?dup | Likely benign |
| SMAD9 | 82 | -261_-256delCGCTGC | Deletion | NM_001127217.2(SMAD9):c.-261_-256delCGCTGC | Likely benign |
| SMAD9 | 85 | -258T>C | single nucleotide variant | NM_001127217.2(SMAD9):c.-258T>C | Benign |
| SMAD9 | 293 | -50G>A | single nucleotide variant | NM_001127217.2(SMAD9):c.-50G>A | Likely benign |
| SMAD9 | 306 | -37C>T | single nucleotide variant | NM_001127217.2(SMAD9):c.-37C>T | Benign |
| SMAD9 | 355 | 12C>T | single nucleotide variant | NM_001127217.2(SMAD9):c.12C>T (p.Thr4=) | Benign |
| SMAD9 | 391 | 48C>T | single nucleotide variant | NM_001127217.2(SMAD9):c.48C>T (p.Pro16=) | Likely benign |
| SMAD9 | 670 | 327G>A | single nucleotide variant | NM_001127217.2(SMAD9):c.327G>A (p.Pro109=) | Likely benign |
| SMAD9 | 691 | 348A>G | single nucleotide variant | NM_001127217.2(SMAD9):c.348A>G (p.Pro116=) | Likely benign |

| | | | | | |
|---|---|---|---|---|---|
| SMAD9 | 777 | 434C>A | single nucleotide variant | NM_001127217.2(SMAD9):c.434C>A (p.Pro145Gln) | Likely benign |
| SMAD9 | 830 | 487G>A | single nucleotide variant | NM_001127217.2(SMAD9):c.487G>A (p.Ala163Thr) | Likely benign |
| SMAD9 | 1086 | 743C>A | single nucleotide variant | NM_001127217.2(SMAD9):c.743C>A (p.Thr248Lys) | Likely benign |
| SMAD9 | 1099 | 756T>C | single nucleotide variant | NM_001127217.2(SMAD9):c.756T>C (p.His252=) | Benign |
| SMAD9 | 1171 | 828C>T | single nucleotide variant | NM_001127217.2(SMAD9):c.828C>T (p.Val276=) | Likely benign |
| SMAD9 | 1347 | 1004-6C>T | single nucleotide variant | NM_001127217.2(SMAD9):c.1004-6C>T | Likely benign |
| SMAD9 | 1378 | 1035G>A | single nucleotide variant | NM_001127217.2(SMAD9):c.1035G>A (p.Val345=) | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*47G>A | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*118C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*200A>C | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*298A>G | Benign |
| SMAD9 | NA | | Duplication | NM_001127217.2(SMAD9):c.*382dupT | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*396A>G | Benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*435delC | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*557T>C | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*573T>C | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*590T>A | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*615A>G | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*637C>T | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*793C>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*850C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*870T>C | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*948A>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*970G>A | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*992G>A | Likely benign |

| | | | | | |
|---|---|---|---|---|---|
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*997C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1031C>T | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1224A>G | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1379G>T | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1430A>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1667A>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*1696G>A | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2036C>A | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2138C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2216G>A | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2333A>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2387A>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2418C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2465T>C | Benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*2506_*2511delTAATCT | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2546A>C | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2653C>T | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2684G>C | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*2905T>C | Likely benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*3171_*3176delAACATT | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3263T>G | Likely benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*3354_*3358delGTTTT | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3543A>G | Likely benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3560T>C | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3562C>T | Benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*3572_*3575delCACA | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3599A>T | Benign |

| | | | | | |
|---|---|---|---|---|---|
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3713T>C | Likely benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*3756_*3759delTTCT | Likely benign |
| SMAD9 | NA | | Deletion | NM_001127217.2(SMAD9):c.*3768_*3770delAAC | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3778C>A | Benign |
| SMAD9 | NA | | single nucleotide variant | NM_001127217.2(SMAD9):c.*3835C>A | Likely benign |
| TBX4 | 61 | 16G>A | single nucleotide variant | NM_018488.3(TBX4):c.16G>A (p.Gly6Ser) | Likely benign |
| TBX4 | 62 | 17G>C | single nucleotide variant | NM_018488.3(TBX4):c.17G>C (p.Gly6Ala) | Benign |
| TBX4 | 149 | 104C>T | single nucleotide variant | NM_018488.3(TBX4):c.104C>T (p.Ala35Val) | Likely benign |
| TBX4 | 149 | c.104C>T (p.A35V) | single nucleotide variant | c.104C>T (p.A35V) | Pathogenic |
| TBX4 | 217 | 172G>A | single nucleotide variant | NM_018488.3(TBX4):c.172G>A (p.Ala58Thr) | Likely benign |
| TBX4 | 232 | 187-15C>T | single nucleotide variant | NM_018488.3(TBX4):c.187-15C>T | Likely benign |
| TBX4 | 274 | c.229T>C(**) | single nucleotide variant | c.229T>C(**) | Pathogenic |
| TBX4 | 274 | c.229T>C, p.W77R | single nucleotide variant | c.229T>C, p.W77R | Pathogenic |
| TBX4 | 294 | 249G>A | single nucleotide variant | NM_018488.3(TBX4):c.249G>A (p.Ala83=) | Likely benign |
| TBX4 | 321 | 276T>G | single nucleotide variant | NM_018488.3(TBX4):c.276T>G (p.Ala92=) | Benign |
| TBX4 | 447 | 402-8G>A | single nucleotide variant | NM_018488.3(TBX4):c.402-8G>A | Benign |
| TBX4 | 667 | 622G>A | single nucleotide variant | NM_018488.3(TBX4):c.622G>A (p.Gly208Ser) | Likely benign |
| TBX4 | 748 | 703-8C>T | single nucleotide variant | NM_018488.3(TBX4):c.703-8C>T | Likely benign |
| TBX4 | 966 | 921C>T | single nucleotide variant | NM_018488.3(TBX4):c.921C>T (p.Asn307=) | Likely benign |
| TBX4 | 977 | 932C>T | single nucleotide variant | NM_018488.3(TBX4):c.932C>T (p.Ser311Leu) | Likely benign |

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|---|---|---|---|---|---|
| TBX4 | 986 | 941C>T | single nucleotide variant | NM_018488.3(TBX4):c.941C>T (p.Ala314Val) | Benign |
| TBX4 | 1050 | 1005C>T | single nucleotide variant | NM_018488.3(TBX4):c.1005C>T (p.His335=) | Likely benign |
| TBX4 | 1128 | 1083G>C | single nucleotide variant | NM_018488.3(TBX4):c.1083G>C (p.Val361=) | Likely benign |
| TBX4 | 1200 | 1155G>A | single nucleotide variant | NM_018488.3(TBX4):c.1155G>A (p.Glu385=) | Likely benign |
| TBX4 | 1259 | 1214C>G | single nucleotide variant | NM_018488.3(TBX4):c.1214C>G (p.Ser405Cys) | Likely benign |
| TBX4 | 1269 | 1224C>T | single nucleotide variant | NM_018488.3(TBX4):c.1224C>T (p.Asp408=) | Benign |
| TBX4 | 1491 | 1446C>T | single nucleotide variant | NM_018488.3(TBX4):c.1446C>T (p.Val482=) | Benign |
| TBX4 | 1557 | 1512G>A | single nucleotide variant | NM_018488.3(TBX4):c.1512G>A (p.Lys504=) | Likely benign |
| TBX4 | 1566 | 1521G>A | single nucleotide variant | NM_018488.3(TBX4):c.1521G>A (p.Ser507=) | Likely benign |
| TBX4 | 1665 | 1620G>A | single nucleotide variant | NM_018488.3(TBX4):c.1620G>A (p.Glu540=) | Likely benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*766A>C | Likely benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*722C>A | Likely benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*707G>T | Benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*327G>A | Benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*313A>C | Likely benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*138A>G | Benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*99T>A | Likely benign |
| TBX4 | NA | | Duplication | NM_018488.3(TBX4):c.*136_*137dupGT | Benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*25G>A | Likely benign |
| TBX4 | NA | | single nucleotide variant | NM_018488.3(TBX4):c.*7C>T | Likely benign |

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|---|---|---|---|---|---|
| THBS1 | 1316 | c.1137G>A p.Asp362Asn | single nucleotide variant | c.1137G>A p.Asp362Asn | Pathogenic |
| THBS1 | 1469 | 1290G>A | single nucleotide variant | NM_003246.3(THBS1):c.1290G>A (p.Lys430=) | Benign |
| THBS1 | 1589 | 1410C>T | single nucleotide variant | NM_003246.3(THBS1):c.1410C>T (p.Asn470=) | Benign |
| THBS1 | 1742 | 1563C>T | single nucleotide variant | NM_003246.3(THBS1):c.1563C>T (p.Asn521=) | Benign |
| THBS1 | 1746 | 1567A>G | single nucleotide variant | NM_003246.3(THBS1):c.1567A>G (p.Thr523Ala) | Benign |
| THBS1 | 2105 | c.1926+255G>A(Intron) | single nucleotide variant | c.1926+255G>A(Intron) | Pathogenic |
| THBS1 | 3047 | 2868T>C | single nucleotide variant | NM_003246.3(THBS1):c.2868T>C (p.Asp956=) | Benign |
| TOPBP1 | 1057 | c.925C>T p.Arg309Cys | single nucleotide variant | c.925C>T p.Arg309Cys | Pathogenic |
| TOPBP1 | 2582 | c.2450C>T p.Ser817Leu | single nucleotide variant | c.2450C>T p.Ser817Leu | Pathogenic |
| TOPBP1 | 3257 | c.3125A>G p.Asn1042Ser | single nucleotide variant | c.3125A>G p.Asn1042Ser | Pathogenic |
| TRPC6 | 64 | -361A>T | single nucleotide variant | NM_004621.5(TRPC6):c.-361A>T | Benign |
| TRPC6 | 65 | -360T>C | single nucleotide variant | NM_004621.5(TRPC6):c.-360T>C | Likely benign |
| TRPC6 | 82 | -343C>G | single nucleotide variant | NM_004621.5(TRPC6):c.-343C>G | Likely benign |
| TRPC6 | 171 | -254C>G | single nucleotide variant | NM_004621.5(TRPC6):c.-254C>G | Benign |
| TRPC6 | 207 | -218C>T | single nucleotide variant | NM_004621.5(TRPC6):c.-218C>T | Benign |
| TRPC6 | 415 | -10C>A | single nucleotide variant | NM_004621.5(TRPC6):c.-10C>A | Likely benign |
| TRPC6 | 426 | c.1-361A>T | single nucleotide variant | c.1-361A>T | Pathogenic |
| TRPC6 | 426 | c.1-218C>T | single nucleotide variant | c.1-218C>T | Pathogenic |
| TRPC6 | 468 | 43C>T | single nucleotide variant | NM_004621.5(TRPC6):c.43C>T (p.Pro15Ser) | Benign |
| TRPC6 | 596 | 171-20A>G | single nucleotide variant | NM_004621.5(TRPC6):c.171-20A>G | Benign |
| TRPC6 | 597 | 172C>T | single nucleotide variant | NM_004621.5(TRPC6):c.172C>T (p.Arg58Trp) | Likely benign |
| TRPC6 | 679 | c. 254 C>G | single nucleotide variant | c. 254 C>G | Pathogenic |
| TRPC6 | 761 | 336A>C | single nucleotide variant | NM_004621.5(TRPC6):c.336A>C (p.Pro112=) | Likely benign |

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|---|---|---|---|---|---|
| TRPC6 | 1313 | 888G>A | single nucleotide variant | NM_004621.5(TRPC6):c.888G>A (p.Thr296=) | Likely benign |
| TRPC6 | 1636 | 1211C>T | single nucleotide variant | NM_004621.5(TRPC6):c.1211C>T (p.Ala404Val) | Benign |
| TRPC6 | 1763 | 1338C>T | single nucleotide variant | NM_004621.5(TRPC6):c.1338C>T (p.His446=) | Likely benign |
| TRPC6 | 1769 | 1344C>A | single nucleotide variant | NM_004621.5(TRPC6):c.1344C>A (p.Ala448=) | Likely benign |
| TRPC6 | 2102 | 1677C>T | single nucleotide variant | NM_004621.5(TRPC6):c.1677C>T (p.Asp559=) | Likely benign |
| TRPC6 | 2108 | 1683T>C | single nucleotide variant | NM_004621.5(TRPC6):c.1683T>C (p.Asn561=) | Benign |
| TRPC6 | 2243 | 1818T>C | single nucleotide variant | NM_004621.5(TRPC6):c.1818T>C (p.Ser606=) | Likely benign |
| TRPC6 | 2513 | 2088C>T | single nucleotide variant | NM_004621.5(TRPC6):c.2088C>T (p.Asn696=) | Likely benign |
| TRPC6 | 2540 | 2115C>T | single nucleotide variant | NM_004621.5(TRPC6):c.2115C>T (p.Tyr705=) | Benign |
| TRPC6 | 2567 | 2142G>A | single nucleotide variant | NM_004621.5(TRPC6):c.2142G>A (p.Thr714=) | Likely benign |
| TRPC6 | 2954 | 2529C>T | single nucleotide variant | NM_004621.5(TRPC6):c.2529C>T (p.Phe843=) | Benign |
| TRPC6 | 3070 | 2645-22_2645-20delCTT | Deletion | NM_004621.5(TRPC6):c.2645-22_2645-20delCTT | Benign |
| TRPC6 | 3137 | 2712G>A | single nucleotide variant | NM_004621.5(TRPC6):c.2712G>A (p.Gln904=) | Benign |
| TRPC6 | 3195 | 2770C>T | single nucleotide variant | NM_004621.5(TRPC6):c.2770C>T (p.Pro924Ser) | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*9C>T | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*13C>T | Likely benign |
| TRPC6 | NA | | Deletion | NM_004621.5(TRPC6):c.*33delT | Likely benign |

| | | | | | |
|---|---|---|---|---|---|
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*40T>G | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*175A>C | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*314C>G | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*336C>T | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*337G>A | Likely benign |
| TRPC6 | NA | | Duplication | NM_004621.5(TRPC6):c.*509_*512dupCTTA | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*530T>C | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*553T>G | Benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*668G>A | Likely benign |
| TRPC6 | NA | | Insertion | NM_004621.5(TRPC6):c.*710_*711insTTTA | Benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*792G>A | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*970A>G | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*1038T>G | Likely benign |
| TRPC6 | NA | | single nucleotide variant | NM_004621.5(TRPC6):c.*1089C>T | Likely benign |

| Phenotype | Exon | Citation (if included in ClinVar) | Mutation ID |
|---|---|---|---|
| not specified | 0 | | ClinVar=212792 |
| not specified | 0 | | ClinVar=136292 |
| Haemorrhagic telangiectasia 2 | 3 | | ClinVar=161202 |
| Osler hemorrhagic telangiectasia syndrome | 3 | | ClinVar=309440 |
| Hereditary hemorrhagic telangiectasia type 2 | 3 | | ClinVar=416700 |
| Osler hemorrhagic telangiectasia syndrome | 3 | | ClinVar=136293 |
| Primary pulmonary hypertension | 3 | PMID: 25543221 | Manual Annotation |
| not specified | 4 | | ClinVar=254710 |
| Hereditary hemorrhagic telangiectasia type 2 | 4 | | ClinVar=212791 |
| Primary pulmonary hypertension | 5 | PMID: 20056902 | Manual Annotation |
| Hereditary hemorrhagic telangiectasia type 2 | 6 | | ClinVar=220940 |
| not specified | 6 | | ClinVar=254711 |
| Hereditary hemorrhagic telangiectasia type 2 | 6 | | ClinVar=212793 |
| not specified | 6 | | ClinVar=389037 |
| not specified | 6 | | ClinVar=254712 |
| Primary pulmonary hypertension | 7 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 7 | PMID: 23298310 | Manual Annotation |
| Hereditary hemorrhagic telangiectasia type 2 | 8 | | ClinVar=416701 |
| Hereditary hemorrhagic telangiectasia type 2 | 8 | | ClinVar=254706 |
| Primary pulmonary hypertension | 8 | PMID: 20056902 | Manual Annotation |

| | | | |
|---|---|---|---|
| Primary pulmonary hypertension | 8 | PMID: 20056902 | Manual Annotation |
| Hereditary hemorrhagic telangiectasia type 2 | 8 | | ClinVar=254708 |
| Hereditary hemorrhagic telangiectasia type 2 | 8 | | ClinVar=254707 |
| not specified | 9 | | ClinVar=379523 |
| Primary pulmonary hypertension | 9 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 9 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 9 | PMID: 20056902 | Manual Annotation |
| not specified | 9 | | ClinVar=254709 |
| Primary pulmonary hypertension | 10 | PMID: 21651515 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 21651515 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 21651515 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 23298310 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 20056902 | Manual Annotation |
| Primary pulmonary hypertension | 10 | PMID: 25543221 | Manual Annotation |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309485 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309484 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309483 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309481 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309480 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309477 |

| | | | |
|---|---|---|---|
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309474 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309470 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309467 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309466 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309465 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309464 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309463 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309462 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309461 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309460 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309459 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309457 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309456 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309455 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309454 |

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| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309453 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309452 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309445 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=309444 |
| Renal dysplasia | 0 | | ClinVar=343666 |
| Renal dysplasia | 4 | | ClinVar=256759 |
| Primary pulmonary hypertension | 4 | PMID: 19332265 | Manual Annotation |
| Renal dysplasia | 4 | | ClinVar=256758 |
| Renal dysplasia | 0 | | ClinVar=369406 |
| Renal dysplasia | 0 | | ClinVar=343686 |
| Renal dysplasia | 0 | | ClinVar=343684 |
| Renal dysplasia | 0 | | ClinVar=343683 |
| Renal dysplasia | 0 | | ClinVar=343680 |
| Renal dysplasia | 0 | | ClinVar=343678 |
| Renal dysplasia | 0 | | ClinVar=343677 |
| Brachydactyly | 4 | | ClinVar=350113 |
| Brachydactyly | 6 | | ClinVar=350115 |
| not specified | 6 | | ClinVar=283921 |
| Brachydactyly | 7 | | ClinVar=218545 |
| Brachydactyly | 7 | | ClinVar=350116 |
| Primary pulmonary hypertension | 8 | PMID: 22374147 | Manual Annotation |
| Primary pulmonary hypertension | 8 | PMID: 22374147 | Manual Annotation |

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| Brachydactyly | 9 | | ClinVar=350117 |
| Brachydactyly | 9 | | ClinVar=350118 |
| Brachydactyly | 10 | | ClinVar=350121 |
| Brachydactyly | 10 | | ClinVar=350122 |
| Brachydactyly | 11 | | ClinVar=350123 |
| Primary pulmonary hypertension | 11 | PMID: 22374147 | Manual Annotation |
| Primary pulmonary hypertension | 11 | PMID: 22374147 | Manual Annotation |
| Brachydactyly | 13 | | ClinVar=350126 |
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| Brachydactyly | 0 | | ClinVar=350182 |
| Brachydactyly | 0 | | ClinVar=350181 |
| Brachydactyly | 0 | | ClinVar=350179 |
| Brachydactyly | 0 | | ClinVar=350178 |
| Brachydactyly | 0 | | ClinVar=350176 |
| Brachydactyly | 0 | | ClinVar=350175 |
| Brachydactyly | 0 | | ClinVar=350174 |
| Brachydactyly | 0 | | ClinVar=350173 |
| Brachydactyly | 0 | | ClinVar=350171 |
| Brachydactyly | 0 | | ClinVar=350169 |
| Brachydactyly | 0 | | ClinVar=350168 |
| Brachydactyly | 0 | | ClinVar=350167 |
| Brachydactyly | 0 | | ClinVar=350166 |
| Brachydactyly | 0 | | ClinVar=350164 |
| Brachydactyly | 0 | | ClinVar=350161 |
| Brachydactyly | 0 | | ClinVar=350160 |
| Brachydactyly | 0 | | ClinVar=350159 |

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| Brachydactyly | 0 | | ClinVar=350157 |
| Brachydactyly | 0 | | ClinVar=350156 |
| Brachydactyly | 0 | | ClinVar=350155 |
| Brachydactyly | 0 | | ClinVar=350154 |
| Brachydactyly | 0 | | ClinVar=350152 |
| Brachydactyly | 0 | | ClinVar=350151 |
| Brachydactyly | 0 | | ClinVar=350149 |
| Brachydactyly | 0 | | ClinVar=350148 |
| Brachydactyly | 0 | | ClinVar=350147 |
| Brachydactyly | 0 | | ClinVar=350146 |
| Brachydactyly | 0 | | ClinVar=350140 |
| Brachydactyly | 0 | | ClinVar=350138 |
| Brachydactyly | 0 | | ClinVar=350137 |
| Brachydactyly | 0 | | ClinVar=350133 |
| Brachydactyly | 0 | | ClinVar=350132 |
| Brachydactyly | 0 | PMID:19738052 | ClinVar=350131 |
| Brachydactyly | 0 | | ClinVar=350129 |
| Brachydactyly | 0 | | ClinVar=350128 |
| Brachydactyly | 0 | | ClinVar=350127 |
| Brachydactyly | 0 | PMID:http://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=4&from=96075833&to=96075833 | ClinVar=287875 |
| Primary pulmonary hypertension | 0 | | ClinVar=333617 |
| Primary pulmonary hypertension | 0 | | ClinVar=333622 |
| Primary pulmonary hypertension | 0 | | ClinVar=333620 |
| Primary pulmonary hypertension | 0 | | ClinVar=333627 |
| Primary pulmonary hypertension | 0 | | ClinVar=333633 |
| Primary pulmonary hypertension | 0 | | ClinVar=333634 |
| Primary pulmonary hypertension | 0 | | ClinVar=333635 |

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| Primary pulmonary hypertension | 0 | | ClinVar=333636 |
| Primary pulmonary hypertension | 2 | | ClinVar=333638 |
| Primary pulmonary hypertension | 5 | | ClinVar=162791 |
| Primary pulmonary hypertension | 5 | | ClinVar=333639 |
| not specified | 5 | | ClinVar=257624 |
| Primary pulmonary hypertension | 6 | | ClinVar=333640 |
| not specified | 7 | | ClinVar=257625 |
| Primary pulmonary hypertension | 12 | | ClinVar=136528 |
| Primary pulmonary hypertension | 12 | | ClinVar=333648 |
| Primary pulmonary hypertension | 12 | | ClinVar=333649 |
| Primary pulmonary hypertension | 12 | | ClinVar=333650 |
| Primary pulmonary hypertension | 12 | | ClinVar=136529 |
| Primary pulmonary hypertension | 13 | | ClinVar=415837 |
| Primary pulmonary hypertension | 0 | | ClinVar=333731 |
| Primary pulmonary hypertension | 0 | | ClinVar=333728 |
| Primary pulmonary hypertension | 0 | | ClinVar=333727 |
| Primary pulmonary hypertension | 0 | | ClinVar=333725 |
| Primary pulmonary hypertension | 0 | | ClinVar=333723 |
| Primary pulmonary hypertension | 0 | | ClinVar=333722 |
| Primary pulmonary hypertension | 0 | | ClinVar=333721 |
| Primary pulmonary hypertension | 0 | | ClinVar=333719 |
| Primary pulmonary hypertension | 0 | | ClinVar=333718 |

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| Primary pulmonary hypertension | 0 | | ClinVar=333717 |
| Primary pulmonary hypertension | 0 | | ClinVar=333716 |
| Primary pulmonary hypertension | 0 | | ClinVar=333715 |
| Primary pulmonary hypertension | 0 | | ClinVar=333713 |
| Primary pulmonary hypertension | 0 | | ClinVar=333709 |
| Primary pulmonary hypertension | 0 | | ClinVar=333708 |
| Primary pulmonary hypertension | 0 | | ClinVar=333705 |
| Primary pulmonary hypertension | 0 | | ClinVar=333703 |
| Primary pulmonary hypertension | 0 | | ClinVar=333701 |
| Primary pulmonary hypertension | 0 | | ClinVar=333700 |
| Primary pulmonary hypertension | 0 | | ClinVar=333697 |
| Primary pulmonary hypertension | 0 | | ClinVar=333695 |
| Primary pulmonary hypertension | 0 | | ClinVar=333694 |
| Primary pulmonary hypertension | 0 | | ClinVar=333688 |
| Primary pulmonary hypertension | 0 | | ClinVar=333687 |
| Primary pulmonary hypertension | 0 | | ClinVar=333685 |
| Primary pulmonary hypertension | 0 | | ClinVar=333684 |
| Primary pulmonary hypertension | 0 | | ClinVar=333682 |
| Primary pulmonary hypertension | 0 | | ClinVar=333680 |
| Primary pulmonary hypertension | 0 | | ClinVar=333679 |
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| Primary pulmonary hypertension | 0 | | ClinVar=333676 |
| Primary pulmonary hypertension | 0 | | ClinVar=333674 |
| Primary pulmonary hypertension | 0 | | ClinVar=333673 |
| Primary pulmonary hypertension | 0 | | ClinVar=333672 |
| Primary pulmonary hypertension | 0 | | ClinVar=333670 |
| Primary pulmonary hypertension | 0 | | ClinVar=333668 |
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| Primary pulmonary hypertension | 0 | | ClinVar=333665 |

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| Primary pulmonary hypertension | 0 | | ClinVar=333664 |
| Primary pulmonary hypertension | 0 | | ClinVar=333660 |
| Primary pulmonary hypertension | 0 | | ClinVar=333659 |
| Primary pulmonary hypertension | 0 | | ClinVar=333657 |
| Primary pulmonary hypertension | 0 | | ClinVar=333656 |
| Primary pulmonary hypertension | 0 | | ClinVar=333655 |
| not specified | 1 | | ClinVar=379541 |
| not specified | 1 | | ClinVar=212818 |
| not specified | 2 | | ClinVar=128610 |
| Primary pulmonary hypertension | 3 | PMID: 22474227 | Manual Annotation |
| Primary pulmonary hypertension | 3 | PMID: 22474227 | Manual Annotation |
| Primary pulmonary hypertension | 3 | PMID: 22474227 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 21537392 | Manual Annotation |
| Primary pulmonary hypertension | 5 | PMID: 21537392 | Manual Annotation |
| Primary pulmonary hypertension | 6 | PMID: 21773759 | Manual Annotation |
| Primary pulmonary hypertension | 6 | PMID: 21773759 | Manual Annotation |
| not specified | 1 | | ClinVar=381178 |
| not specified | 9 | | ClinVar=381213 |
| not specified | 11 | | ClinVar=381179 |
| not specified | 12 | | ClinVar=402821 |
| not specified | 12 | | ClinVar=381180 |
| Primary pulmonary hypertension | 23 | PMID: 25512148 | Manual Annotation |
| not specified | 25 | | ClinVar=381181 |

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| not specified | 28 | | ClinVar=381182 |
| not specified | 31 | | ClinVar=381183 |
| not specified | 31 | | ClinVar=381184 |
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| not specified | 0 | | ClinVar=402822 |
| not specified | 0 | | ClinVar=381187 |
| not specified | 0 | | ClinVar=381186 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=365100 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=365097 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=414302 |
| Osler hemorrhagic telangiectasia syndrome | 1 | | ClinVar=178774 |
| Primary pulmonary hypertension | 1 | PMID: 15687131 | Manual Annotation |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=414303 |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=237016 |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=237019 |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=237021 |

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| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=414311 |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=137208 |
| not specified | 2 | | ClinVar=255144 |
| Osler hemorrhagic telangiectasia syndrome | 2 | | ClinVar=237026 |
| Osler hemorrhagic telangiectasia syndrome | 3 | | ClinVar=414305 |
| Osler hemorrhagic telangiectasia syndrome | 3 | | ClinVar=414306 |
| Osler hemorrhagic telangiectasia syndrome | 3 | | ClinVar=386058 |
| not specified | 3 | | ClinVar=255145 |
| Osler hemorrhagic telangiectasia syndrome | 4 | | ClinVar=414312 |
| not specified | 4 | | ClinVar=213199 |
| Primary pulmonary hypertension | 4 | PMID: 25543221 | Manual Annotation |
| Osler hemorrhagic telangiectasia syndrome | 5 | | ClinVar=365092 |
| Osler hemorrhagic telangiectasia syndrome | 5 | | ClinVar=213200 |
| Primary pulmonary hypertension | 5 | PMID: xx | Manual Annotation |
| Osler hemorrhagic telangiectasia syndrome | 6 | | ClinVar=414313 |
| Osler hemorrhagic telangiectasia syndrome | 7 | | ClinVar=237031 |
| Osler hemorrhagic telangiectasia syndrome | 7 | | ClinVar=365090 |

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|---|---|---|---|
| not specified | 7 | | ClinVar=213201 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=414301 |
| not specified | 8 | | ClinVar=387433 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=414308 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=137209 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=137210 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=365089 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=163406 |
| Osler hemorrhagic telangiectasia syndrome | 8 | | ClinVar=414307 |
| Osler hemorrhagic telangiectasia syndrome | 10 | | ClinVar=237017 |
| Osler hemorrhagic telangiectasia syndrome | 11 | | ClinVar=213202 |
| Osler hemorrhagic telangiectasia syndrome | 12 | | ClinVar=414309 |
| Osler hemorrhagic telangiectasia syndrome | 12 | | ClinVar=414304 |
| Osler hemorrhagic telangiectasia syndrome | 12 | | ClinVar=255142 |
| not specified | 12 | | ClinVar=385734 |

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| Osler hemorrhagic telangiectasia syndrome | 12 | | ClinVar=237020 |
| Primary pulmonary hypertension | 12 | PMID: 26167679 | Manual Annotation |
| not specified | 13 | | ClinVar=388615 |
| not specified | 13 | | ClinVar=255143 |
| Osler hemorrhagic telangiectasia syndrome | 13 | | ClinVar=414310 |
| Osler hemorrhagic telangiectasia syndrome | 14 | | ClinVar=213203 |
| Osler hemorrhagic telangiectasia syndrome | 14 | | ClinVar=161229 |
| Osler hemorrhagic telangiectasia syndrome | 14 | | ClinVar=237025 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=365078 |
| Osler hemorrhagic telangiectasia syndrome | 0 | | ClinVar=226632 |
| Familial atrial fibrillation | 0 | | ClinVar=309329 |
| not provided | 1 | | ClinVar=309332 |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 17267549 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 17267549 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 23861362 | Manual Annotation |
| Familial atrial fibrillation | 1 | | ClinVar=309336 |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 17267549 | Manual Annotation |

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| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 17267549 | Manual Annotation |
| not specified | 1 | | ClinVar=191570 |
| not specified | 1 | | ClinVar=191571 |
| not specified | 1 | | ClinVar=258594 |
| Primary pulmonary hypertension | 1 | PMID: 25189502 | Manual Annotation |
| not specified | 1 | | ClinVar=191573 |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 24936649 | Manual Annotation |
| Familial atrial fibrillation | 0 | | ClinVar=369020 |
| Familial atrial fibrillation | 0 | | ClinVar=309343 |
| Familial atrial fibrillation | 0 | | ClinVar=309342 |
| Primary pulmonary hypertension | 1 | PMID: 23883380 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23883380 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23883380 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23883380 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23883380 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23883380 | Manual Annotation |
| not specified | 0 | | ClinVar=402999 |
| Primary pulmonary hypertension | 5 | PMID: 24439467 | Manual Annotation |
| not specified | 2 | | ClinVar=256119 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 3 | | ClinVar=256131 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 3 | | ClinVar=328419 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 4 | PMID:24086431 | ClinVar=328418 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 4 | | ClinVar=256148 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 4 | | ClinVar=328417 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 6 | | ClinVar=328414 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 6 | | ClinVar=328413 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 7 | | ClinVar=256117 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 7 | | ClinVar=256118 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 9 | | ClinVar=256120 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 9 | PMID:20038773 | ClinVar=256121 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 11 | | ClinVar=328410 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 11 | | ClinVar=328409 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 11 | | ClinVar=256122 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 11 | | ClinVar=256123 |
| not specified | 11 | | ClinVar=256124 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 13 | | ClinVar=256125 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 14 | | ClinVar=256126 |
| not specified | 15 | | ClinVar=256127 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 16 | | ClinVar=256128 |
| Primary pulmonary hypertension | 16 | PMID: 24936512 | Manual Annotation |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 16 | | ClinVar=256129 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 17 | | ClinVar=328405 |

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| Primary pulmonary hypertension | 17 | PMID: 24936512 | Manual Annotation |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 17 | | ClinVar=256130 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 21 | | ClinVar=256132 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 22 | | ClinVar=328396 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 22 | | ClinVar=328395 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 22 | | ClinVar=256133 |
| not specified | 22 | | ClinVar=256134 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 22 | | ClinVar=328393 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 23 | | ClinVar=328392 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 24 | | ClinVar=328390 |
| not specified | 24 | | ClinVar=256136 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 24 | | ClinVar=328388 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 25 | | ClinVar=328385 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 25 | | ClinVar=256137 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 25 | | ClinVar=256138 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 25 | | ClinVar=256139 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 29 | | ClinVar=256140 |
| not specified | 30 | | ClinVar=256141 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=256142 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=328383 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=256143 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=328382 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=328381 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 30 | | ClinVar=256144 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 31 | | ClinVar=328379 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 32 | | ClinVar=256145 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 32 | | ClinVar=256146 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256147 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256149 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=328374 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256150 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256151 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=328372 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=328371 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256152 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256153 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 33 | | ClinVar=256154 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328370 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328369 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328368 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328366 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328364 |

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| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328363 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328362 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328361 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328360 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328359 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328357 |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | 0 | | ClinVar=328356 |
| Plasminogen activator inhibitor type 1 deficiency | 2 | | ClinVar=358307 |
| Plasminogen activator inhibitor type 1 deficiency | 4 | | ClinVar=358311 |
| Primary pulmonary hypertension | 4 | PMID: 20300292 | Manual Annotation |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358338 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358333 |

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| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358329 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358325 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358323 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358320 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358319 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358318 |
| Plasminogen activator inhibitor type 1 deficiency | 0 | | ClinVar=358316 |
| Primary pulmonary hypertension | 3 | PMID: 25284742 | Manual Annotation |
| not specified | 0 | | ClinVar=392894 |
| not specified | 0 | | ClinVar=388624 |
| not specified | 0 | | ClinVar=420335 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224033 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224026 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224023 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224021 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224016 |
| not specified | 0 | | ClinVar=382523 |

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| Juvenile polyposis syndrome | 0 | | ClinVar=240139 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224029 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224027 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224022 |
| Hereditary cancer-predisposing syndrome | 0 | | ClinVar=224020 |
| not specified | 0 | | ClinVar=422097 |
| not specified | 0 | | ClinVar=390388 |
| not specified | 0 | | ClinVar=389873 |
| not specified | 0 | | ClinVar=380201 |
| not specified | 0 | | ClinVar=385850 |
| Juvenile polyposis syndrome | 2 | | ClinVar=183887 |
| Primary pulmonary hypertension | 2 | PMID: 21898662 | Manual Annotation |
| not specified | 2 | | ClinVar=378972 |
| Hereditary cancer-predisposing syndrome | 2 | | ClinVar=233358 |
| Juvenile polyposis syndrome | 2 | | ClinVar=183699 |
| not specified | 2 | | ClinVar=388467 |
| Primary pulmonary hypertension | 2 | PMID: 21898662 | Manual Annotation |
| Juvenile polyposis syndrome | 2 | | ClinVar=382652 |

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| Juvenile polyposis syndrome | 2 | | ClinVar=240147 |
| Juvenile polyposis syndrome | 2 | | ClinVar=240148 |
| Juvenile polyposis syndrome | 2 | | ClinVar=132767 |
| Hereditary cancer-predisposing syndrome | 2 | | ClinVar=183926 |
| Juvenile polyposis syndrome | 2 | | ClinVar=372022 |
| not specified | 3 | | ClinVar=378623 |
| Hereditary cancer-predisposing syndrome | 3 | | ClinVar=224025 |
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| Hereditary cancer-predisposing syndrome | 3 | | ClinVar=183782 |
| Juvenile polyposis syndrome | 3 | | ClinVar=240149 |
| Myhre syndrome | 3 | PMID:16436638 | ClinVar=24799 |
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| Juvenile polyposis syndrome | 3 | | ClinVar=230174 |
| Juvenile polyposis syndrome | 3 | | ClinVar=215843 |
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| Juvenile polyposis syndrome | 3 | | ClinVar=413433 |
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| not specified | 3 | | ClinVar=391022 |
| Hereditary cancer-predisposing syndrome | 3 | | ClinVar=224037 |
| Hereditary cancer-predisposing syndrome | 4 | | ClinVar=224032 |
| Juvenile polyposis syndrome | 4 | | ClinVar=219454 |
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| not specified | 5 | | ClinVar=259750 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=224034 |
| Myhre syndrome | 5 | | ClinVar=139218 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=185721 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=183859 |
| not specified | 5 | | ClinVar=391104 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=231885 |
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| Juvenile polyposis syndrome | 5 | | ClinVar=380411 |
| Juvenile polyposis syndrome | 5 | | ClinVar=220122 |

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| Juvenile polyposis syndrome | 5 | | ClinVar=413438 |
| Juvenile polyposis syndrome | 5 | | ClinVar=240151 |
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| not specified | 5 | | ClinVar=380355 |
| Juvenile polyposis syndrome | 5 | | ClinVar=378625 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=224028 |
| Hereditary cancer-predisposing syndrome | 5 | | ClinVar=224019 |
| Juvenile polyposis syndrome | 6 | | ClinVar=413441 |
| Hereditary cancer-predisposing syndrome | 6 | | ClinVar=224017 |
| Juvenile polyposis syndrome | 6 | | ClinVar=215844 |
| not specified | 6 | | ClinVar=378626 |
| not specified | 6 | | ClinVar=224018 |
| not specified | 7 | | ClinVar=420249 |
| Hereditary cancer-predisposing syndrome | 7 | | ClinVar=233821 |
| Juvenile polyposis syndrome | 7 | | ClinVar=240154 |
| Juvenile polyposis syndrome | 7 | | ClinVar=413442 |
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| Hereditary cancer-predisposing syndrome | 7 | | ClinVar=224024 |
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| Juvenile polyposis syndrome | 8 | PMID:Unpublished ARUP data | ClinVar=24817 |
| Juvenile polyposis syndrome | 8 | PMID:16287957 | ClinVar=24797 |
| Juvenile polyposis syndrome | 8 | | ClinVar=413439 |
| Hereditary cancer-predisposing syndrome | 8 | | ClinVar=233198 |
| Juvenile polyposis syndrome | 8 | | ClinVar=220195 |
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| Myhre syndrome | 9 | | ClinVar=132693 |
| Juvenile polyposis syndrome | 9 | | ClinVar=413431 |
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| Juvenile polyposis syndrome | 10 | | ClinVar=413435 |
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| not specified | 10 | | ClinVar=381470 |
| Juvenile polyposis syndrome | 10 | | ClinVar=139219 |

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| Juvenile polyposis syndrome | 10 | | ClinVar=240144 |
| Juvenile polyposis syndrome | 10 | | ClinVar=231979 |
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| Juvenile polyposis syndrome | 10 | | ClinVar=413432 |
| not specified | 10 | | ClinVar=384825 |
| Hereditary cancer-predisposing syndrome | 10 | | ClinVar=224036 |
| Hereditary cancer-predisposing syndrome | 10 | | ClinVar=224035 |
| Hereditary cancer-predisposing syndrome | 10 | | ClinVar=224031 |
| Juvenile polyposis syndrome | 11 | | ClinVar=413434 |
| Juvenile polyposis syndrome | 11 | | ClinVar=413440 |
| Juvenile polyposis syndrome | 11 | | ClinVar=132745 |
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| Juvenile polyposis syndrome | 11 | | ClinVar=184070 |
| Juvenile polyposis syndrome | 11 | | ClinVar=240145 |
| Hereditary cancer-predisposing syndrome | 12 | | ClinVar=224030 |

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| Primary pulmonary hypertension | 12 | PMID: 21898662 | Manual Annotation |
| Juvenile polyposis syndrome | 12 | | ClinVar=184561 |
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| Hereditary cancer-predisposing syndrome | 12 | | ClinVar=233281 |
| Juvenile polyposis syndrome | 12 | | ClinVar=136073 |
| Juvenile polyposis syndrome | 12 | | ClinVar=378975 |
| Juvenile polyposis syndrome | 12 | | ClinVar=413437 |
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| Juvenile polyposis syndrome | 12 | | ClinVar=136074 |
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| Myhre syndrome | 0 | | ClinVar=327218 |
| Myhre syndrome | 0 | | ClinVar=327217 |
| Myhre syndrome | 0 | | ClinVar=327215 |
| Myhre syndrome | 0 | | ClinVar=327212 |

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| Myhre syndrome | 0 | | ClinVar=327208 |
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| Myhre syndrome | 0 | | ClinVar=327141 |

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| Myhre syndrome | 0 | | ClinVar=327126 |
| Myhre syndrome | 0 | | ClinVar=327125 |
| Myhre syndrome | 0 | | ClinVar=327123 |
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| Myhre syndrome | 0 | | ClinVar=139220 |
| Juvenile polyposis syndrome | 0 | | ClinVar=136071 |
| Primary pulmonary hypertension | 0 | | ClinVar=311917 |
| Primary pulmonary hypertension | 0 | | ClinVar=311919 |
| Primary pulmonary hypertension | 0 | | ClinVar=311908 |
| Primary pulmonary hypertension | 0 | | ClinVar=213809 |
| Primary pulmonary hypertension | 2 | | ClinVar=227076 |
| Primary pulmonary hypertension | 2 | | ClinVar=227954 |
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| Primary pulmonary hypertension | 2 | | ClinVar=311905 |

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| Primary pulmonary hypertension | 3 | | ClinVar=311904 |
| Primary pulmonary hypertension | 3 | | ClinVar=213810 |
| Primary pulmonary hypertension | 4 | | ClinVar=311902 |
| Primary pulmonary hypertension | 4 | | ClinVar=227077 |
| Primary pulmonary hypertension | 5 | | ClinVar=311901 |
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| Primary pulmonary hypertension | 6 | | ClinVar=311897 |
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| Primary pulmonary hypertension | 0 | | ClinVar=311890 |
| Primary pulmonary hypertension | 0 | | ClinVar=311889 |
| Primary pulmonary hypertension | 0 | | ClinVar=311888 |
| Primary pulmonary hypertension | 0 | | ClinVar=311885 |
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| Primary pulmonary hypertension | 0 | | ClinVar=311883 |
| Primary pulmonary hypertension | 0 | | ClinVar=311881 |
| Primary pulmonary hypertension | 0 | | ClinVar=311880 |
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| Primary pulmonary hypertension | 0 | | ClinVar=311868 |
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| Primary pulmonary hypertension | 0 | | ClinVar=311865 |

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| Primary pulmonary hypertension | 0 | | ClinVar=311864 |
| Primary pulmonary hypertension | 0 | | ClinVar=311863 |
| Primary pulmonary hypertension | 0 | | ClinVar=311860 |
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| Primary pulmonary hypertension | 0 | | ClinVar=311829 |
| Primary pulmonary hypertension | 0 | | ClinVar=311828 |
| Primary pulmonary hypertension | 0 | | ClinVar=311826 |
| Primary pulmonary hypertension | 0 | | ClinVar=311825 |
| Primary pulmonary hypertension | 0 | | ClinVar=311821 |
| Primary pulmonary hypertension | 0 | | ClinVar=311817 |
| Primary pulmonary hypertension | 0 | | ClinVar=311814 |

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| Primary pulmonary hypertension | 0 | | ClinVar=311813 |
| Primary pulmonary hypertension | 0 | | ClinVar=311811 |
| Primary pulmonary hypertension | 0 | | ClinVar=311810 |
| Primary pulmonary hypertension | 0 | | ClinVar=311809 |
| Primary pulmonary hypertension | 0 | | ClinVar=311807 |
| Ischiopatellar dysplasia | 1 | | ClinVar=324240 |
| Ischiopatellar dysplasia | 1 | | ClinVar=324241 |
| Ischiopatellar dysplasia | 1 | | ClinVar=324243 |
| Primary pulmonary hypertension | 1 | PMID: 23592887 | Manual Annotation |
| Ischiopatellar dysplasia | 1 | | ClinVar=324246 |
| Ischiopatellar dysplasia | 2 | | ClinVar=324247 |
| Primary pulmonary hypertension | 2 | PMID: 23592887 | Manual Annotation |
| Primary pulmonary hypertension | 2 | PMID: 23592887 | Manual Annotation |
| Ischiopatellar dysplasia | 2 | | ClinVar=324248 |
| Ischiopatellar dysplasia | 2 | | ClinVar=324249 |
| Ischiopatellar dysplasia | 4 | | ClinVar=261034 |
| Ischiopatellar dysplasia | 5 | | ClinVar=324251 |
| Ischiopatellar dysplasia | 6 | | ClinVar=324252 |
| Ischiopatellar dysplasia | 7 | | ClinVar=324255 |
| Ischiopatellar dysplasia | 7 | | ClinVar=324256 |

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| Ischiopatellar dysplasia | 7 | | ClinVar=261035 |
| Ischiopatellar dysplasia | 7 | | ClinVar=324257 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324259 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324260 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324261 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324262 |
| Ischiopatellar dysplasia | 8 | | ClinVar=261033 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324263 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324264 |
| Ischiopatellar dysplasia | 8 | | ClinVar=324265 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324286 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324285 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324284 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324280 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324279 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324277 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324274 |
| Ischiopatellar dysplasia | 0 | | ClinVar=324268 |
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| Ischiopatellar dysplasia | 0 | | ClinVar=324266 |

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| Primary pulmonary hypertension | 8 | PMID: 22198906 | Manual Annotation |
| not specified | 8 | | ClinVar=403534 |
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| not specified | 10 | | ClinVar=403536 |
| not specified | 10 | | ClinVar=403537 |
| Primary pulmonary hypertension | 12 | PMID: 22198906 | Manual Annotation |
| not specified | 18 | | ClinVar=403538 |
| Primary pulmonary hypertension | 8 | PMID: 24702692 | Manual Annotation |
| Primary pulmonary hypertension | 14 | PMID: 24702692 | Manual Annotation |
| Primary pulmonary hypertension | 19 | PMID: 24702692 | Manual Annotation |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301926 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301925 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301923 |
| Focal segmental glomerulosclerosis | 0 | PMID:19380626 | ClinVar=301920 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301918 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301914 |
| Primary pulmonary hypertension | 1 | PMID: 19380626 | Manual Annotation |
| Primary pulmonary hypertension | 1 | PMID: 19380626 | Manual Annotation |
| Focal segmental glomerulosclerosis | 1 | | ClinVar=259463 |
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| Focal segmental glomerulosclerosis | 2 | | ClinVar=301913 |
| Primary pulmonary hypertension | 2 | PMID: 19380626 | Manual Annotation |
| Focal segmental glomerulosclerosis | 2 | | ClinVar=301911 |

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| Focal segmental glomerulosclerosis | 4 | | ClinVar=259454 |
| Focal segmental glomerulosclerosis | 5 | | ClinVar=301908 |
| Focal segmental glomerulosclerosis | 5 | | ClinVar=301907 |
| Focal segmental glomerulosclerosis | 6 | | ClinVar=301906 |
| Focal segmental glomerulosclerosis | 6 | | ClinVar=259455 |
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| Focal segmental glomerulosclerosis | 8 | | ClinVar=301902 |
| Focal segmental glomerulosclerosis | 8 | | ClinVar=259458 |
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| Focal segmental glomerulosclerosis | 11 | | ClinVar=259460 |
| not specified | 13 | | ClinVar=259461 |
| Focal segmental glomerulosclerosis | 13 | | ClinVar=259462 |
| Focal segmental glomerulosclerosis | 13 | | ClinVar=301898 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301897 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301896 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301895 |

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| Focal segmental glomerulosclerosis | 0 | | ClinVar=301894 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301893 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301892 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301891 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301890 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301889 |
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| Focal segmental glomerulosclerosis | 0 | | ClinVar=301887 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301885 |
| Focal segmental glomerulosclerosis | 0 | | ClinVar=301884 |
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| Focal segmental glomerulosclerosis | 0 | | ClinVar=301881 |
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| Focal segmental glomerulosclerosis | 0 | | ClinVar=301878 |