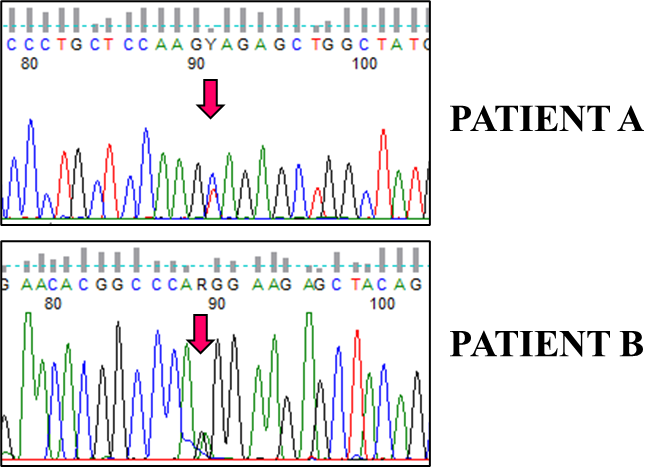
****

**Supplemental Figure 1**: **TREX1 electropherograms of patients A and B.** The chromatograms show the presence of c.407C>T (p.A136V) (upper panel) and c.520A>G (p.R174G) (lower panel) variants in patient A and B, respectively.

Diagram

Description automatically generated

**Supplemental Figure 2:** **Structural context of hTREX1’s mutated residues.** A structure of mTREX11-242 bound to ssDNA (PDB = ‘2IOC’) was used as a template with MODELLER v9.22 to model the hTREX11-242 structure with the originally disordered loops resolved. The respective protomers of the hTREX1 homodimer are shown as dark and light grey cartoons, 4-mer oligos (‘DNA’) are shown as blue sticks, divalent metal ions are shown as black spheres, A136 residues are shown as red sticks, and R174 residues are shown as green sticks. Graphics were generated with PyMOL v2.3.2 (The PyMOL Molecular Graphics System, Version 2.0 Schrödinger, LLC), and figure was prepared in PowerPoint.

Graphical user interface

Description automatically generated

**Supplemental Figure 3: dsDNA degradation by A136V and R174G variants.** **[a,b]** *Agarose Gel Visualization of TREX1 Exonuclease Activity*. Standard exonuclease reactions were prepared with equimolar concentrations of the indicated enzymes, incubated at room temperature for 0, 5, 10, 20, or 40 minutes, quenched in ethanol, then visualized on 0.8% agarose gels. Supercoiled dsDNA plasmid is nicked to generate initial substrate (‘Nicked), and TREX1 degrades one strand of the substrate over time to generate ssDNA plasmid (‘ssDNA’). **[c,d]** *Quantification of Agarose Gels*. Band intensity for each of the 0-minute lanes on the gels in panels a-b was quantified by densitometry, along with band intensity on the corresponding region of the associated lanes. Differences in band intensity were normalized to the 0-minute lanes’ band, and these values (‘Relative Degradation’) were plotted as a function of time. Gels and plots are representative of two separate experiments.

Graphical user interface

Description automatically generated

**Supplemental Figure 4: ssDNA degradation by A136V and R174G variants.** **[a,b]** *Fluorescence-Based Quantification of TREX1 dsDNA Degradation*. Standard exonuclease reactions were prepared with equimolar concentrations of the indicated enzymes, incubated at room temperature for the indicated times, quenched in QuantiFluor ssDNA dye, and ssDNA content measured by fluorescence. Plots of fluorescence vs time were generated and fit with one-phase decay nonlinear regression in Prism 7.0 (GraphPad). Plots are composites of 18 different reactions from 3 different experiments. Data points indicate mean, and error bars represent standard deviation. **[c]** *Activity Rates of TREX1 Variants*. Initial velocities were quantified from the respective regression lines in panels a-b and normalized to wild-type initial velocity to calculate relative activity. Values are mean and standard deviation. ‘[S] >> Km’ refers to ~5 µM of a 30-mer oligo.

Graphical user interface

Description automatically generated

**Supplemental Figure 5: Quality control data from A136V and R174G preps.** **[a,c]** *Chromatography Purification of hT1A136V/A136V (panel a) & hT1R174G/R174G (panel c).* Samples were taken at the indicated steps of chromatography purification and visualized by SDS-PAGE with Coomassie staining. (1) pre-induction culture sample, (2) post-induction culture sample, (3) nickel column applicant, (4) nickel column wash, (5) nickel column eluent, (6) post-cleavage p-cell column applicant, (7) p-cell column wash, & (8) p-cell column eluent. **[b,d]** *Chromatography Purification of hT1A136V/+ (panel b) & hT1R174G /+ (panel d).* (1) pre-induction culture sample, (2) post-induction culture sample, (3) amylose column applicant, (4) amylose column wash, (5) amylose column eluent / nickel column applicant, (6) nickel column wash, (7) nickel column eluent, (8) post-cleavage p-cell column applicant, (9) p-cell column wash, & (10) p-cell column eluent. **[e]** *Validation of Enzyme Purities and Concentrations.* Protein content of preps was visualized via SDS-PAGE with Coomassie staining. Enzyme preps were used to load 0.75 µg each, as measured by A280, onto a 12% gel. Band intensities were quantified using GelAnalyzer 19.1, then plotted as mean and range. Graph was prepared with Prism 7.0 (GraphPad), and figure was assembled in PowerPoint.

**Supplemental Table 1:** genes responsible for Mendelian conditions causing monogenic forms of stroke.

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **GENE** | **OMIM# gene** | **OMIM# disease** | **inheritance pattern** | **GenBank ID** | **coding exons** | **target submitted (bp)** | **target designed (bp)** | **missed (bp)** | **covered (%)** | **Reference**  **(PMID)** |
| ABCA1 | 600046 | 604091  205400 | AD  AR | NM\_005502 | 49 | 7276 | 7276 | 0 | 100 | 30278532 |
| ABCC6 | 603234 | 614473  264800  177850 | AR  AR  AD | NM\_001171 | 31 | 4903 | 4755 | 148 | 96,98 | 24008425  29709427  29722917 |
| ABCD4 | 603214 | 614857 | AR | NM\_005050 | 19 | 2771 | 2771 | 0 | 100 | 20301503 |
| ABCG5 | 605459 | 618666 | AR | NM\_022436 | 13 | 2606 | 2606 | 0 | 100 | 11099417 |
| ABCG8 | 605460 | 210250 | AR | NM\_022437 | 13 | 2672 | 2672 | 0 | 100 | 11099417 |
| ACP5 | 171640 | 607944 | AR | NM\_001111036 | 4 | 1178 | 1178 | 0 | 100 | 26951490 |
| ACTA2 | 102620 | 611788  613834 | AD | NM\_001613 | 8 | 1214 | 1214 | 0 | 100 | 19409525  30661495 |
| ACTN1 | 102575 | 615193 | AD | NM\_001102 | 22 | 3845 | 3845 | 0 | 100 | 30356112 |
| ACVRL1 | 601284 | 600376 | AD | NM\_000020 | 9 | 1602 | 1602 | 0 | 100 | 31637968 |
| ADAMTS13 | 604134 | 274150 | AR | NM\_139025 | 29 | 5734 | 5722 | 12 | 99,79 | 30356112 |
| ADAR | 146920 | 615010 | AR | NM\_001111 | 15 | 4431 | 4431 | 0 | 100 | 25672750 |
| ALG8 | 608103 | 608104 | AR | NM\_024079 | 14 | 2336 | 2231 | 105 | 95,5 | 12480927 |
| ANO6 | 608663 | 262890 | AR | NM\_001025356 | 23 | 4226 | 4226 | 0 | 100 | 30356112 |
| APP | 104760 | 605714 | AD | NM\_201413 | 19 | 2545 | 2545 | 0 | 100 | 7131028 |
| ATP7A | 300011 | 309400 | XLR | NM\_000052.7 | 22 | 4723 | 4723 | 0 | 100 | 11118799 |
| BGN | 301870 | 300989 | XL | NM\_001711.6 | 7 | 1457 | 1457 | 0 | 100 | 27632686 |
| BRAF | 164757 | 115150  613707  613706 | AD | NM\_001374258.1 | 18 | 2481 | 2481 | 0 | 100 | 12187019 |
| BRCC3 | 300617 | 300845 | XLR | NM\_001018055.3 | 11 | 1504 | 1504 | 0 | 100 | 25733922 |
| C1R | 613785 | 130080 | AD | NM\_001733.7 | 9 | 2336 | 2336 | 0 | 100 | 25485215 |
| CBS | 613381 | 236800 | AR | NM\_000071.3 | 15 | 1806 | 1806 | 0 | 100 | 26689889 |
| CCER2 | 617634 | - | - | NM\_001243212 | 5 | 851 | 851 | 0 | 100 | 27717682 |
| CCM1 | 604214 | 116860 | AD | NM\_004912 | 16 | 2851 | 2851 | 0 | 100 | 9811928 |
| CCM2 | 607929 | 603284 | AD | NM\_031443.4 | 11 | 1868 | 1868 | 0 | 100 | 9811928 |
| CD59 | 107271 | 612300 | AR | NM\_000611 | 3 | 537 | 537 | 0 | 100 | 25716358 |
| CECR1 | 607575 | 182410  615688 | AR | NM\_001282225.2 | 10 | 2066 | 2066 | 0 | 100 | 24552284 |
| COG6 | 606977 | 614576 | AR | NM\_020751.3 | 20 | 2996 | 2996 | 0 | 100 | 20605848 |
| COL1A1 | 120150 | 130060 | AD | NM\_000088.4 | 51 | 4905 | 4905 | 0 | 100 | 28089253  12695216 |
| COL1A2 | 120160 | 617821  225320 | AD  AR | NM\_000089.4 | 52 | 4621 | 4621 | 0 | 100 | 28089253  12695216 |
| COL3A1 | 120180 | 130050  618343 | AD  AR | NM\_000090.4 | 51 | 4911 | 4911 | 0 | 100 | 12786757 |
| COL4A1 | 120130 | 611773  175780  618564 | AD | NM\_001845.6 | 53 | 5564 | 5564 | 0 | 100 | 30413629 |
| COL4A2 | 120090 | 614483 | AD | NM\_001846.4 | 47 | 7489 | 7489 | 0 | 100 | 30413629 |
| COL4A3 | 120070 | 203780  104200 | AR  AD | NM\_000091 | 52 | 5533 | 5533 | 0 | 100 | 10893738 |
| COL4A4 | 120131 | 203780 | AD | NM\_000092 | 47 | 7423 | 7423 | 0 | 100 | 10893738 |
| COL4A5 | 303630 | 301050 | XLD | NM\_000495 | 51 | 5573 | 5573 | 0 | 100 | 30045277 |
| COL4A6 | 303631 | - | - | NM\_001847 | 47 | 7488 | 7488 | 0 | 100 | 30045277 |
| COL5A1 | 120215 | 130000 | AD | NM\_000093 | 67 | 6256 | 6256 | 0 | 100 | 17053184 |
| COL5A2 | 120190 | 130010 | AD | NM\_000393 | 54 | 7200 | 7200 | 0 | 100 | 17053184 |
| CST3 | 604312 | 105150 | AD | NM\_000099 | 3 | 591 | 591 | 0 | 100 | 3472718 |
| CTSA | 613111 | - | AD | NM\_000308 | 15 | 2247 | 2247 | 0 | 100 | 27664989 |
| CYP11B1 | 610613 | 202010  103900 | AR  AD | NM\_000497 | 9 | 1602 | 1602 | 0 | 100 | 10852446 |
| DPM1 | 603503 | 608799 | AR | NM\_003859 | 10 | 1388 | 1321 | 67 | 95,1 | 15669674 |
| DYRK1B | 604556 | 615812 | AD | NM\_004714 | 10 | 2390 | 2390 | 0 | 100 | 24827035 |
| ELN | 130160 | 123700  185500 | AD | NM\_001278939 | 34 | 4094 | 4094 | 0 | 100 | 18348261 |
| ENG | 131195 | 187300 | AD | NM\_000118 | 15 | 2753 | 2753 | 0 | 100 | 27352867  31637968 |
| ENPP1 | 173335 | 208000  615522  613312 | AR  AD  AR | NM\_006208 | 25 | 4028 | 4028 | 0 | 100 | 19206175 |
| EPAS1 | 603349 | 611783 | AD | NM\_001430 | 16 | 3413 | 3413 | 0 | 100 | 18184961 |
| EPHB4 | 600011 | 618196 | AD | NM\_004444 | 17 | 3134 | 3134 | 0 | 100 | 28687708 |
| EPOR | 133171 | 133100 | AD | NM\_000121 | 8 | 1927 | 1927 | 0 | 100 | 3371213 |
| ESCO2 | 609353 | - | - | NM\_001017420 | 10 | 2306 | 2306 | 0 | 100 | 1642282 |
| F10 | 613872 | 227600 | AR | NM\_000504 | 8 | 1867 | 1867 | 0 | 100 | 3732313 |
| F13A1 | 134570 | 613225 | AR | NM\_000129 | 14 | 2899 | 2899 | 0 | 100 | 9550516 |
| F13B | 134580 | 613235 | AR | NM\_001994 | 12 | 2586 | 2586 | 0 | 100 | 30356112 |
| F2 | 176930 | 613679  188050 | AR  AD | NM\_000506 | 14 | 2009 | 2009 | 0 | 100 | 28160964 |
| F5 | 612309 | 227400  188055 | AR  AD | NM\_000130 | 25 | 6925 | 6925 | 0 | 100 | 30635457 |
| F7 | 613878 | 227500 | AR | NM\_000131 | 9 | 1761 | 1761 | 0 | 100 | 30356112 |
| F8 | 300841 | 306700 | XLR | NM\_000132 | 27 | 7350 | 7350 | 0 | 100 | 30356112 |
| F9 | 300746 | 306900  300807 | XLR | NM\_000133 | 8 | 1466 | 1466 | 0 | 100 | 19846852 |
| FBLN4 | 604633 | 614437 | AR | NM\_016938 | 10 | 1432 | 1432 | 0 | 100 | 8985490 |
| FBLN5 | 604580 | 614434  219100  608895 | AD  AR  AD | NM\_006329 | 11 | 1897 | 1897 | 0 | 100 | 8985490 |
| FBN1 | 134797 | 604308  154700 | AD | NM\_000138 | 65 | 9266 | 9266 | 0 | 100 | 29030048 |
| FGA | 134820 | 202400  616004 | AR  AD/AR | NM\_021871 | 6 | 2705 | 2705 | 0 | 100 | 19598064 |
| FGB | 134830 | 202400  616400 | AR  AD/AR | NM\_005141 | 8 | 1876 | 1876 | 0 | 100 | 19598064 |
| FGG | 134850 | 202400  616400 | AR  AD/AR | NM\_000509 | 10 | 1877 | 1877 | 0 | 100 | 19598064 |
| FLI1 | 193067 | 617443 | AD  AR | NM\_001271010 | 10 | 1491 | 1491 | 0 | 100 | 30356112 |
| FLNA | 300017 | 300049  314400 | XLD  XL | NM\_001110556 | 47 | 10294 | 10294 | 0 | 100 | 9883725 |
| FOXC1 | 601090 | 602482 | AD | NM\_001453 | 1 | 1672 | 1672 | 0 | 100 | 25250569 |
| GAA | 606800 | 232300 | AR | NM\_001079803 | 19 | 3049 | 3049 | 0 | 100 | 3322184 |
| GCDH | 608801 | 231670 | AR | NM\_000159 | 12 | 1481 | 1484 | 0 | 100 | 28411331 |
| GDF2 | 605120 | 615506 | AD | NM\_016204 | 2 | 1390 | 1390 | 0 | 100 | 31637968 |
| GFI1B | 604383 | 187900 | AD  AR | NM\_004188 | 6 | 1293 | 1293 | 0 | 100 | 30356112 |
| GGCX | 137167 | 610842  277450 | digenic  AR | NM\_000821 | 15 | 3053 | 3053 | 0 | 100 | 12384421 |
| GLA | 300644 | 301500 | XL | NM\_000169 | 7 | 1360 | 1360 | 0 | 100 | 33835733 |
| GP1BA | 606672 | 231200  153670  177820 | AR  AD  AD | NM\_000173 | 1 | 2009 | 2009 | 0 | 100 | 30356112 |
| GP1BB | 138720 | 231200 | AR | NM\_000407 | 2 | 641 | 641 | 0 | 100 | 30356112 |
| GP6 | 605546 | 614201 | AR | NM\_016363 | 8 | 2263 | 2263 | 0 | 100 | 30356112 |
| GP9 | 173515 | 231200 | AR | NM\_000174 | 1 | 584 | 584 | 0 | 100 | 30356112 |
| GUCY1A3 | 139396 | 615750 | AR | NM\_000856 | 8 | 2157 | 2157 | 0 | 100 | 25733922 |
| HBB | 141900 | 603903 | AR | NM\_000518 | 3 | 474 | 474 | 0 | 100 | 7782612 |
| HRG | 142640 | 613116 | AD | NM\_000412 | 7 | 1928 | 1928 | 0 | 100 | 29108964  8475479 |
| HTRA1 | 602194 | 600142  616779 | AR  AD | NM\_002775 | 9 | 1893 | 1893 | 0 | 100 | 32719647 |
| IFIH1 | 606951 | 615846 | AD | NM\_022168 | 16 | 3878 | 3878 | 0 | 100 | 175395 |
| ITGA2 | 192974 | - | - | NM\_002203 | 30 | 5046 | 5046 | 0 | 100 | 30356112 |
| ITGA2B | 607759 | 187800 | AD | NM\_000419 | 30 | 4620 | 4620 | 0 | 100 | 30356112 |
| ITGB3 | 173470 | 619271 | AD | NM\_000212 | 15 | 2517 | 2517 | 0 | 100 | 30356112 |
| ITM2B | 603904 | 176500  117300 | AD | NM\_021999 | 6 | 1101 | 1101 | 0 | 100 | 10391242 |
| IVD | 607036 | 243500 | AR | NM\_002225 | 12 | 1401 | 1401 | 0 | 100 | 20142522 |
| JAG1 | 601920 | 118450 | AD | NM\_000214 | 26 | 3917 | 3917 | 0 | 100 | 14993126 |
| JAK2 | 147796 | 614521 | AD | NM\_004972 | 23 | 3629 | 3629 | 0 | 100 | 22397670 |
| JAM3 | 606871 | 613730 | AR | NM\_032801 | 9 | 1383 | 1383 | 0 | 100 | 21109224 |
| KNG1 | 612358 | 228960 | AR | NM\_000893 | 11 | 2566 | 2566 | 0 | 100 | 12576314 |
| KRAS | 190070 | 609942 | AD | NM\_004985 | 5 | 737 | 737 | 0 | 100 | 12187019 |
| LDLR | 606945 | 143890 | AD/AR | NM\_001195799 | 18 | 2763 | 2763 | 0 | 100 | 1301956 |
| LDLRAP1 | 605747 | 603813 | AR | NM\_015627 | 9 | 1377 | 1377 | 0 | 100 | 9626156 |
| LMAN1 | 601567 | 227300 | AR | NM\_005570 | 13 | 2183 | 2183 | 0 | 100 | 30356112 |
| LMBRD1 | 612625 | 277380 | AR | NM\_018368 | 16 | 2423 | 2423 | 0 | 100 | 20301503 |
| LMNA | 150330 | 115200 | AD | NM\_170707 | 15 | 2369 | 2369 | 0 | 100 | 19095983 |
| LOX | 153455 | 617168 | AD | NM\_001178102 | 7 | 1604 | 1604 | 0 | 100 | 27432961 |
| LZTR1 | 600574 | 616564  605275 | AD  AR | NM\_006767 | 21 | 2733 | 2733 | 0 | 100 | 12187019 |
| MCFD2 | 607788 | 613625 | AR | NM\_001171507 | 4 | 733 | 733 | 0 | 100 | 30356112 |
| MFAP5 | 601103 | 616166 | AD | NM\_003480 | 9 | 972 | 972 | 0 | 100 | 25434006 |
| MGAT2 | 602616 | 212066 | AR | NM\_002408 | 1 | 1394 | 1394 | 0 | 100 | 19419693  1159665 |
| MMACHC | 609831 | 277400 | AR | NM\_015506 | 4 | 889 | 889 | 0 | 100 | 18986243 |
| MMADHC | 611935 | 277410 | AR | NM\_015702 | 7 | 961 | 961 | 0 | 100 | 20301409 |
| MPI | 154550 | 602579 | AR | NM\_002435 | 8 | 1672 | 1672 | 0 | 100 | 10484808 |
| MPL | 159530 | 601977  604498 | AD  AR | NM\_005373 | 12 | 2028 | 2028 | 0 | 100 | 15269348 |
| MTHFR | 607093 | 236250 | AR | NM\_005957 | 11 | 2081 | 2081 | 0 | 100 | 26839351  29849257 |
| MTRR | 602568 | 236270 | AR | NM\_002454 | 15 | 2328 | 2279 | 49 | 97,9 | 29849257 |
| MTR | 156570 | 250940 | AR | NM\_000254 | 33 | 4128 | 4128 | 0 | 100 | 29849257 |
| MYH11 | 160745 | 132900 | AD | NM\_002474 | 42 | 6391 | 6391 | 0 | 100 | 30661495 |
| MYLK | 600922 | 613780 | AD | NM\_053025 | 31 | 6055 | 6055 | 0 | 100 | 29544503  26147384 |
| NBEAL2 | 614169 | 139090 | AR | NM\_015175 | 54 | 10965 | 10965 | 0 | 100 | 30356112 |
| NF1 | 613113 | 162200  601321 | AD | NM\_001042492 | 58 | 9161 | 9161 | 0 | 100 | 30661495 |
| NOTCH1 | 190198 | 616028  109730 | AD | NM\_017617 | 34 | 8008 | 7987 | 21 | 99,7 | 25132448  9631276 |
| NOTCH3 | 600276 | 125310 | AD | NM\_000435 | 33 | 7296 | 7296 | 0 | 100 | 19539236 |
| NRAS | 164790 | 613224 | AD | NM\_002524 | 4 | 610 | 610 | 0 | 100 | 12187019 |
| P2RY12 | 600515 | 609821 | AR | NM\_022788 | 1 | 1079 | 1079 | 0 | 100 | 30356112 |
| PCCA | 232000 | 606054 | AR | NM\_000282 | 24 | 2427 | 2427 | 0 | 100 | 18986243 |
| PCCB | 232050 | 606054 | AR | NM\_000532 | 16 | 2480 | 2480 | 0 | 100 | 18986243 |
| PCNT | 605925 | 210720 | AR | NM\_006031 | 47 | 12361 | 12361 | 0 | 100 | 34016138 |
| PDCD10 | 609118 | 603285 | AD | NM\_007217 | 7 | 989 | 989 | 0 | 100 | 17041941 |
| PDE3A | 123805 | 112410 | AD | NM\_000921 | 17 | 4321 | 4293 | 28 | 99,3 | 25961942 |
| PDE4D | 600129 | 614613 | AD | NM\_001104631 | 22 | 4450 | 4450 | 0 | 100 | 22464250 |
| PGM1 | 171900 | 614921 | AR | NM\_002633 | 12 | 2589 | 2589 | 0 | 100 | 10484808 |
| PITX2 | 601542 | 180500 | AD | NM\_153426 | 5 | 1209 | 1209 | 0 | 100 | 25250569 |
| PKD1 | 601313 | 173900 | AD | NM\_001009944 | 46 | 13372 | 13278 | 94 | 99,3 | 8130364  27567292 |
| PKD2 | 173910 | 613095 | AD | NM\_000297 | 15 | 3057 | 3057 | 0 | 100 | 27567292 |
| PLAU | 191840 | 601709 | AD | NM\_002658 | 11 | 1880 | 1880 | 0 | 100 | 30356112 |
| PLG | 173350 | 217090 | AR | NM\_000301 | 19 | 2627 | 2627 | 0 | 100 | 8392398 |
| PLOD1 | 153454 | 225400 | AR | NM\_000302 | 20 | 3325 | 3325 | 0 | 100 | 9617436 |
| PLOD3 | 603066 | 612394 | AR | NM\_001084 | 19 | 3167 | 3167 | 0 | 100 | 18834968 |
| PMM2 | 601785 | 212065 | AR | NM\_000303 | 8 | 1141 | 1141 | 0 | 100 | 29470411 |
| POLR3F | 617455 | - | AD | NM\_006466 | 9 | 1041 | 1041 | 0 | 100 | 30115567 |
| PRDX1 | 176763 | 277400 | AR | NM\_181697 | 5 | 650 | 650 | 0 | 100 | 29302025 |
| PRKACG | 176893 | 616176 | AR | NM\_002732 | 1 | 1066 | 1066 | 0 | 100 | 30356112 |
| PRKG1 | 176894 | 615436 | AD | NM\_001098512 | 19 | 3087 | 3087 | 0 | 100 | 23910461 |
| PROC | 612283 | 176860  612304 | AD  AR | NM\_000312 | 8 | 1466 | 1466 | 0 | 100 | 20187890  1511989 |
| PROCR | 600646 | - | AD | NM\_006404 | 4 | 757 | 757 | 0 | 100 | 11552992 |
| PROS1 | 176880 | 612336  614514 | AD  AR | NM\_000313 | 16 | 2287 | 2287 | 0 | 100 | 20484936 |
| PTPN11 | 176876 | 163950 | AD | NM\_002834 | 15 | 1936 | 1936 | 0 | 100 | 12187019 |
| RAF1 | 164760 | 611553 | AD | NM\_002880 | 16 | 2107 | 2107 | 0 | 100 | 12187019 |
| RANBP2 | 602752 | - | - | NM\_006267 | 29 | 9965 | 8963 | 1002 | 89,9 | 29593631 |
| RASA1 | 139150 | 608354 | AD | NM\_002890 | 26 | 3412 | 3411 | 1 | 99,9 | 24038909 |
| RASGRP2 | 605577 | 615888 | AR | NM\_001098671 | 15 | 2580 | 2580 | 0 | 100 | 30356112 |
| RIT1 | 609591 | 615355 | AD | NM\_001256821 | 6 | 771 | 771 | 0 | 100 | 12187019 |
| RNASEH2A | 606034 | 610333 | AR | NM\_006397 | 8 | 1300 | 1300 | 0 | 100 | 32642802 |
| RNASEH2B | 610326 | 610181 | AR | NM\_024570 | 12 | 1572 | 1572 | 0 | 100 | 32642802 |
| RNASEH2C | 610330 | 610329 | AR | NM\_032193 | 4 | 695 | 695 | 0 | 100 | 32642802 |
| RNF213 | 613768 | 607151 | AD  AR | NM\_001256071 | 67 | 16462 | 16439 | 23 | 99,8 | 31650369  26147384 |
| SAG | 181031 | 258100 | AR | NM\_000541 | 15 | 1968 | 1968 | 0 | 100 | 22665972 |
| SAMHD1 | 606754 | 612952 | AR | NM\_015474 | 16 | 2681 | 2681 | 0 | 100 | 20842748 |
| SCN5A | 600163 | 614022 | AD | NM\_001099404 | 28 | 6423 | 6423 | 0 | 100 | 16684018 |
| SERPINC1 | 107300 | 613118 | AD  AR | NM\_000488 | 7 | 1465 | 1465 | 0 | 100 | 14347873 |
| SERPIND1 | 142360 | 612356 | AD | NM\_000185 | 4 | 1540 | 1540 | 0 | 100 | 14347873 |
| SERPINE1 | 173360 | 613329 | AD  AR | NM\_000602 | 8 | 1609 | 1609 | 0 | 100 | 1435917 |
| SKI | 164780 | 182212 | AD | NM\_003036 | 7 | 2257 | 2217 | 40 | 98,2 | 24357594 |
| SLC19A2 | 603941 | 249270 | AR | NM\_006996 | 6 | 1794 | 1794 | 0 | 100 | 10720020 |
| SLC20A2 | 158378 | 213600 | AD | NM\_006749 | 10 | 2459 | 2459 | 0 | 100 | 30637044 |
| SLC2A10 | 606145 | 208050 | AR | NM\_030777 | 5 | 1876 | 1876 | 0 | 100 | 17485657 |
| SLFN14 | 614958 | 616913 | AD | NM\_001129820 | 4 | 2779 | 2779 | 0 | 100 | 30356112 |
| SMAD3 | 603109 | 613795 | AD | NM\_005902 | 10 | 1452 | 1452 | 0 | 100 | 21778426 |
| SMAD4 | 600993 | 175050 | AD | NM\_005359 | 11 | 1769 | 1769 | 0 | 100 | 31637968 |
| SMARCAL1 | 606622 | 242900 | AR | NM\_014140 | 16 | 3665 | 3665 | 0 | 100 | 16840568 |
| SOS1 | 182530 | 610733 | AD | NM\_005633 | 23 | 4232 | 4232 | 0 | 100 | 12187019 |
| SOS2 | 601247 | 616559 | AD | NM\_006939 | 23 | 4919 | 4919 | 0 | 100 | 12187019 |
| SPARC | 182120 | 616507 | AR | NM\_003118 | 9 | 1478 | 1478 | 0 | 100 | 26027498 |
| STAT1 | 600555 | 614162 | AD | NM\_007315 | 23 | 2487 | 2487 | 0 | 100 | 28161409 |
| TBXA2R | 188070 | 614009 | AD | NM\_001060 | 3 | 1423 | 1245 | 178 | 87,5 | 30356112 |
| TBXAS1 | 274180 | 614158 | AD | NM\_001130966 | 14 | 2472 | 2472 | 0 | 100 | 30356112 |
| TEK | 600221 | 600195 | AD | NM\_000459 | 23 | 4525 | 4525 | 0 | 100 | 19888299 |
| TGFB2 | 190220 | 614816 | AD | NM\_003238 | 8 | 1409 | 1409 | 0 | 100 | 25835445 |
| TGFB3 | 190230 | 615582 | AD | NM\_003239 | 7 | 1589 | 1589 | 0 | 100 | 25835445 |
| TGFBR1 | 190181 | 609192 | AD | NM\_004612 | 9 | 1614 | 1614 | 0 | 100 | 15731757 |
| TGFBR2 | 190182 | 600168 | AD | NM\_003242 | 8 | 1859 | 1859 | 0 | 100 | 22772368 |
| THBD | 188040 | 614486 | AD | NM\_000361 | 1 | 1738 | 1738 | 0 | 100 | 26354877 |
| THPO | 600044 | 187950 | AD | NM\_000460 | 6 | 1782 | 1782 | 0 | 100 | 9425899 |
| TREX1 | 606609 | 225750  192315 | AD/AR  AD | NM\_033629 | 1 | 1160 | 1160 | 0 | 100 | 11438888 |
| VHL | 608537 | 263400 | AR | NM\_000551 | 3 | 672 | 672 | 0 | 100 | 14726398 |
| VKORC1 | 608547 | 607473 | AR | NM\_024006 | 4 | 776 | 776 | 0 | 100 | 30356112 |
| VWF | 613160 | 193400  277480  613554 | AD  AR  AD/AR | NM\_000552.5 | 51 | 8952 | 08952 | 0 | 100 | 30356112 |
| XYLT1 | 608124 | 264800 | AR | NM\_022166.4 | 12 | 3480 | 3477 | 3 | 99,9 | 16571645 |
| XYLT2 | 608125 | 264800 | AR | NM\_022167.4 | 11 | 3148 | 3148 | 0 | 100 | 16571645 |
| YY1AP1 | 607860 | 602531 | AR | NM\_001198903 | 11 | 2844 | 2844 | 0 | 100 | 27939641 |

Legend: AD=autosomal dominant; AR=autosomal recessive; XL=X-linked; bp=base pairs

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