|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **Gezonde functie** |  |  |  |  |  | **Gen** |  |  | **Intron / Exon** | **Genvariant** |  |  |
|  |  |  |  |  |  | HUGO | NCBI |  |  | GERMONLINE |  |  | puntmutatie |
|  |  |  |  |  |  | algemeen | 1, 2 (T=U) | 3 | 4 |  | 3 |  | 4 |
| RF | Symbool(ccds) | Frame | # basen  | stop | Naam  | Bron | FASTA DNAnucleotidenseqentie | FASTAmRNA | FASTA amino- zuur (expressie-product) |  | FASTA mRNA | **MUTATIE****(in amino-zuur seq.)****1 lettercode****3 lettercode** | FASTA amino-zuur (expressie-product) |
| 1 | hemoglobine B | +3 | 444 | TAA | HBB | [HGNC:4827](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=4827) | [NC 000011.8](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000011.8&from=5203272&to=5204877&strand=2&dopt=fasta) | [NM 000518.4](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_000518.4&dopt=fasta) | [NP 000509.1](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_000509.1&dopt=fasta) | [ENST00000292901](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000292901) | [Sickle cel anemia](http://srs.ebi.ac.uk/srsbin/cgi-bin/wgetz?%5blibs_EQ_%7bembl_SP_emblann%7d-AccNumber:AF059180%5d+-e) | E7VGlu7Val | [VAR\_002863.html](http://web.expasy.org/variant_pages/VAR_002863.html) |
| 2 | glucosylceramidase | +3 | 1611 | TGA | GBA | [HGNC:4177](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=4177) | [NC 000001.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000001.9&from=153470867&to=153481112&strand=2&dopt=fasta) |  |  | [ENST00000327247](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000327247) | Gaucher disease | V433LVal433Leu | [VAR\_003310.html](http://web.expasy.org/variant_pages/VAR_003310.html)  |
| 3 | Factor 8 | +2 | 7056 | TGA | F8 | [HGNC:3546](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3546) | [NG 005114.1](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NG_005114.1&from=498&to=187430&strand=2&dopt=fasta) |  |  | [ENST00000330287](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000330287) | hemofilie A | E340KGlu340Lys | [VAR\_028519.html](http://web.expasy.org/variant_pages/VAR_028519.html)  |
| 4 | chlorideion transport | +2 | 4443 | TAG | CFTR | [HGNC:1884](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=1884) | [NC 000007.12](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000007.12&from=116907253&to=117095955&dopt=fasta) |  |  | [ENST00000003084](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000003084) | taaislijmziekte (cystic fibrosis) | H949YHis949Tyr | [VAR\_000225.html](http://web.expasy.org/variant_pages/VAR_000225.html)  |
| 5 | Kopertransporteiwit | +2 | 4398 | TGA | ATP7B | [HGNC:870](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=870) | [NC 000013.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000013.9&from=51404806&to=51483631&strand=2&dopt=fasta) |  |  | [ENST00000242839](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000242839) | Ziekte van Wilson | G1111DGly1111Asp | [VAR\_023032.html](http://web.expasy.org/variant_pages/VAR_023032.html)  |
| 6 | verbindings eiwit | +3 | 681 | TAA | GJB2 | [HGNC:4284](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=4284) | [NC 000013.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000013.9&from=19659609&to=19665037&strand=2&dopt=fasta) |  |  | [ENST00000382844](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000382844) | KID syndroom(doofheid) |  | [VAR\_015456](http://web.expasy.org/variant_pages/VAR_015456.html) |
| 7 | (mogelijk) genregulator eiwit | +1 | 2787 | TGA | RB1 | [HGNC:9884](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=9884) | [NC000013.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000013.9&from=47775912&to=47954023&dopt=fasta) |  |  | [ENST00000267163](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000267163) | retinoblastoma | R500GArg500Gly | [VAR\_011580.html](http://web.expasy.org/variant_pages/VAR_011580.html)  |
| 8 | Leptine | +1 | 504 | TGA | LEP | [HGNC:6553](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=6553) | [NC\_000007.12](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000007.12&from=127668567&to=127684917&dopt=fasta) | [NM 000230.1](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_000230.1&dopt=fasta) |  | [ENST00000308868](http://www.ensembl.org/Homo_sapiens/exonview?panel_exons=on;db=core;transcript=ENST00000308868) | obesitas | R105WArg105Trp | [VAR\_008094.html](http://web.expasy.org/variant_pages/VAR_008094.html) |
| 9 | DNA reparatie eiwit | +3 | 5375 | TAA | [BRCA-1](http://www.ncbi.nlm.nih.gov/CCDS/CcdsBrowse.cgi?REQUEST=PROTACC&DATA=NP_009228&BUILDS=CURRENTBUILDS) | [HGNC:1100](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=HGNC:1100) | [NC 000017.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000017.9&from=38449840&to=38530994&strand=2&dopt=fasta) | [NM\_007294.2](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&qty=1&c_start=1&list_uids=NM_007294&uids=&dopt=fasta&dispmax=5&sendto=&fmt_mask=0&from=begin&to=end&extrafeatpresent=1&ef_STS=64&ef_CDD=8&ef_MGC=16&ef_HPRD=32&ef_tRNA=128&ef_microRNA=256) | [NP\_009228.1](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_009228.1&dopt=fasta) | [ENST00000303391](http://www.ensembl.org/Homo_sapiens/exonview?db=core;transcript=ENST00000303391) | borstkanker | M1775RMet1775ArgNB: beide az sequenties incl. deleties: leerlingen vinden M1479R of M1704R. Volgend jaar weglaten!! | <http://web.expasy.org/variant_pages/VAR_007799.html>  |
| 10 | Chromosomaal proteine | +3 | 1461 | TGA | MECP2 | [HGNC:6990](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=6990) | [NC 000023.9](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000023.9&from=152940458&to=153016323&strand=2&dopt=fasta) |  |  | [ENST00000303391](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000303391) | Rett Syndroom |  | [VAR\_023553](http://web.expasy.org/variant_pages/VAR_023553.html) |
| 11 | Sulfaat trasport eiwit | +1 | 2220 | TAA | SLC26A2 | [HGNC: 10994](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=10994) | [NC\_000005.8](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000005.8&from=149320639&to=149347156&dopt=fasta) |  |  | [ENST00000286298](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000286298) | dysplasie |  | [VAR\_007437](http://web.expasy.org/variant_pages/VAR_007437.html) |
| 12 | Celoppervlak Iontransport regulatiue eiwit | +2 | 1773 | TGA | NF2 | [HGNC: 7773](http://www.gene.ucl.ac.uk/nomenclature/data/get_data.php?hgnc_id=7773) | [NC\_000022.98](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000022.9&from=28329565&to=28424587&dopt=fasta) | [NM\_181832](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_181832.1&dopt=fasta) |  | [ENST00000334961](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000334961) | neurofibromatose |  | [VAR\_000810](http://web.expasy.org/variant_pages/VAR_000810.html) |
| 13 | Natrium onafhankelijk transporteiwit van chloride en iodide. | +1 | 2343 | TGA | SLC26A4 | [HGNC:8818](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=8818) | [NC\_000007.12](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000007.12&from=107088316&to=107145490&dopt=fasta) |  |  | [ENST00000265715](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000265715) | Pendred syndrome (gehoor verlies) | E29QGlu29Gln | [VAR\_021640.html](http://web.expasy.org/variant_pages/VAR_021640.html)  |
| 14 | Katalyseert de aanzet tot beschikbaar maken van glucose voor glycogeen synthese | +3 | 1398 | TGA | GCK | [HGNC:4195](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=4195) | [NC\_000007.12](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000007.12&from=44150395&to=44195563&strand=2&dopt=fasta) |  |  | [ENST00000336642](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000336642) | Diabetes II, hyperglycemie |  | [VAR\_012352](http://web.expasy.org/variant_pages/VAR_012352.html) |
| 15 | Eiwit, betrokken bij transcriptie | +3 | 1191 | TGA | ERCC8 | [HGNC:3439](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3439) | [NC\_000005.8](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000005.8&from=60205415&to=60276648&strand=2&dopt=fasta) |  |  | [ENST00000265038](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000265038) | Cockayne syndrome (dwerggroei) |  | [VAR\_025380](http://web.expasy.org/variant_pages/VAR_025380.html) |
| 16 | apolipoprotein E (betrokken bij catabolisme lipoproteinen) | +1 | 954 | TGA | APOE | [HGN C:613](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=613) | [NC\_000019.8](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NC_000019.8&from=50100879&to=50104490&dopt=fasta) |  |  | [ENST00000252486](http://www.germonline.org/Homo_sapiens/exonview?db=core;transcript=ENST00000252486) | hyperlipoproteinemia type III / (alzheimer) |  | [VAR\_000656](http://web.expasy.org/variant_pages/VAR_000656.html) |
| 17 | Transcriptie factor | +2 | 615 | TAG | SRY | [HGNC:11311](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=11311) |  |  |  |  | XY vrouwen Swyer syndrome |  | [VAR\_003727](http://web.expasy.org/variant_pages/VAR_003727.html)  |
| 18 | EGF9groeifactor)-receptor | +1 | 1935 | TGA | EGFR | [HGNC:3236](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3236) |  |  |  |  | Longkanker |  | [VAR\_026098](http://web.expasy.org/variant_pages/VAR_026098.html) |
| 19 | ‘Herhaal eiwit’, co-eiwit van RNA polymerase | +2 | 1191 | TGA | ERCC8 | [HGNC:3439](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3439) |  |  |  |  | Cockayne syndroom |  | [VAR\_025380](http://web.expasy.org/variant_pages/VAR_025380.html) |
| 20 | Speelt een vruciale rol in de prenatale ontwikkeling  | +2 | 1212 | TGA | PAX3 | [HGNC:8617](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=8617) |  |  |  |  | Waardenburg syndroom |  | [VAR\_013641](http://web.expasy.org/variant_pages/VAR_013641.html) |
| 21 | Glycoproteine in been en kraakbeen  | +1 | 8955 | TAA | FBN1 | [HGNC:3603](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3603) |  |  |  |  | Marfan synfroom | C832YCys832TyrNB: De 1ste 113 codons van de ORF worden blijkbaar NIET vertaald; lln vinden ook C945Y | <http://web.expasy.org/variant_pages/VAR_023871.html>  |
| 22 | Transcriptiefactor die de apoptose controleert | +1 | 1182 | TGA | TP53 | [HGNC:11998](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=11998) |  |  |  |  | +/- 50% van alle kankers |  | [VAR\_005899](http://web.expasy.org/variant_pages/VAR_005899.html) |
| 23 | transcriptiefactor | +2 | 522 | TGA | CDKN2A | [HGNC:1787](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=1787) |  |  |  |  | +/- 25% van alle kankers |  | [VAR\_029287](http://web.expasy.org/variant_pages/VAR_029287.html) |
| 24 | transcriptiefactor | +1 | 1299 | TGA | WT-1 | [HGNC:12796](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=12796) |  |  |  |  | Wilms (nier) tumor |  | [VAR\_007740](http://web.expasy.org/variant_pages/VAR_007740.html) |
| 25 | Regulator van de beta-cateninefunctie | +1 | 8532 | TAA | APC | [HGNC:583](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=583) |  |  |  | [ENSG00000134982](http://www.ensembl.org/Homo_sapiens/geneview?gene=ENSG00000134982) | Colorectale tumoren |  | [VAR\_005037](http://web.expasy.org/variant_pages/VAR_005037.html) |
| 26 | Tumor supressor eiwit  | +2 | 1848 | TGA | MEN1 | [HGNC:7010](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=7010) |  |  |  |  | Endocriene tumoren (pancreas, bijschildklier) |  | [VAR\_005461](http://web.expasy.org/variant_pages/VAR_005461.html) |
| 27 | Serine, threonine proteïne fosfatase | +1 | 2004 | TAA | PPP2R1B | [HGNC:9303](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=9303) |  |  |  |  | Longkanker en colorectale tumoren |  | [VAR\_022905](http://web.expasy.org/variant_pages/VAR_022905.html)/[VAR\_022904](http://web.expasy.org/variant_pages/VAR_022904.html) |
| 28 | Transmembraan eiwit voor cel-cel adhesie | +2 | 2649 | TAG | CDH1 | [HGNC:1748](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=1748) |  |  |  |  | Maag- en bortskanker |  | [VAR\_001311](http://web.expasy.org/variant_pages/VAR_001131.html) |
| 29 | Coagulatiefactor XIII | +3 | 2199 | TGA | F13A1 | [HGNC:3531](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3531) |  |  |  |  | Deficient factor VIII |  | [VAR\_007474](http://web.expasy.org/variant_pages/VAR_007474.html) |
| 30 | Celregulatieeiwit | +3 | 2979 | TAG | EVC | [HGNC:3497](http://www.ncbi.nlm.nih.gov/entrez/utils/fref.fcgi?http://www.gene.ucl.ac.uk/cgi-bin/nomenclature/get_data.pl?hgnc_id=3497) |  |  |  |  | Ellis van crefeld syndroom |  | [VAR\_009944](http://web.expasy.org/variant_pages/VAR_009944.html) |