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Supplementary Discussion

Supplementary Notes: Base-calling and indel-identification Matlab scripts.

Supplementary Sequences: Primer, gene, and protein sequences used in this work.

Supplementary Table 1: Activities of BE1, BE2, and BE3 at EMX1 off-targets.

Supplementary Table 2: Activities of BE1, BE2, and BE3 at FANCF off-targets.

Supplementary Table 3: Activities of BE1, BE2, and BE3 at HEK293 site 2 off-targets.

Supplementary Table 4: Activities of BE1, BE2, and BE3 at HEK293 site 3 off-targets.

Supplementary Table 5: Activities of BE1, BE2, and BE3 at HEK293 site 4 off-targets.

Supplementary Table 6: Indel formation following treatment of HEK293T cells with BE1, BE2, BE3, and wt Cas9 + a single-stranded DNA template for HDR.

Supplementary Table 7: Mutation rates of non-protospacer bases following BE3-mediated correction of the Alzheimer's disease-associated *APOE4* allele to *APOE3r* in mouse astrocytes.

Supplementary Table 8: Mutation rates of non-protospacer bases following BE3-mediated correction of the cancer-associated p53 Y163C mutation in HCC1954 human cells.

Supplementary Table 9: List of base-editable gene variants associated with human disease containing only a single C within the activity window and an NGG PAM positioned appropriately

Supplementary References

Supplementary Figure 1: Source Gels

SUPPLEMENTARY DISCUSSION

Recent progress to increase HDR and suppress NHEJ. A small-molecule inhibitor of ligase IV, an essential enzyme in the NHEJ pathway, has been shown to increase HDR efficiency.[35,36] However, this strategy is challenging in post-mitotic cells, which typically down-regulate HDR, and its therapeutic relevance is limited by the potential risks of inhibiting ligase IV in non-target cells. Enhanced HDR efficiency can also be achieved by the timed delivery of Cas9-guide RNA complexes into chemically synchronized cells, as HDR efficiency is highly cell-cycle dependent.[37] Such an approach, however, is limited to research applications in cell culture since synchronizing cells is highly disruptive. In some cases, it is possible to design HDR templates such that the product of successful HDR contains mutations in the PAM sequence and therefore is no longer a substrate for subsequent Cas9 modification, increasing the overall yield of HDR products,[38] although such an approach imposes constraints on the product sequences. Recently, this strategy has been coupled to the use of ssDNA donors that are complementary to the non-target strand and high-efficiency ribonucleoprotein (RNP) delivery to substantially increase the efficiency of HDR, but even in these cases the ratio of HDR to NHEJ outcomes is relatively low (< 2).[39]

BE1 activity window. We observed some base editing outside of the typical window of positions 4 to 8 when the substrate C is preceded by a T, which we attribute to the unusually high activity of APOBEC1 for TC substrates.[40]

Testing BE1 in human cells. To test base editing in human cells, we optimized BE1 codon usage for mammalian expression and appended a C-terminal nuclear localization signal (NLS).[41]

Indels in BE1- and BE2-treated cells. Indels will on rare occasion arise from the processing of U:G lesions by cellular repair processes, which involve single-strand break intermediates that are known to lead to indels.[42] Given that several hundred endogenous U:G lesions are generated every day per human cell from spontaneous cytosine deamination,[43] we anticipate that the total indel frequency from U:G lesion repair is unlikely to increase from BE1 or BE2 activity at a single target locus.

Permanence of base editing in cells. We observed no substantial change in editing efficiency between non-passaged HEK293T cells (editing observed in 1.8% to 2.6% of sequenced strands

for the three target Cs with BE2, and 6.2% to 14.3% with BE3) and cells that had undergone approximately five cell divisions after base editing (editing observed in 1.9% to 2.3% of sequenced strands for the same target Cs with BE2, and 6.4% to 14.5% with BE3), confirming that base edits in these cells are durable (Extended Data Fig. 6).

Off-target activity. The off-target activities of Cas9, dCas9, and Cas9 nickase have been extensively studied.[44-47] In general, off-target C to T conversions by BE1, BE2, and BE3 paralleled off-target Cas9 nuclease-mediated genome modification frequencies.

Determination of clinically relevant human genetic diseases treatable by BE2 and BE3. To illuminate the potential relevance of base editors to address human genetic diseases, we searched the NCBI ClinVar database[48] for known genetic diseases that could in principle be corrected by this approach. We filtered ClinVar by first examining only single nucleotide polymorphisms (SNPs), then removing any non-pathogenic variants. Out of the 24,670 pathogenic SNPs, 3,956 are caused by either a T to C, or an A to G, substitution. This list was further filtered to only include variants with a nearby NGG PAM that would position the SNP within the deamination activity window, resulting in 911 clinically relevant pathogenic gene variants that could in principle be corrected by the base editors described here. Of these, 284 contain only one C within the base editing activity window. A detailed list of these pathogenic mutations can be found in Supplementary Table 9.

SUPPLEMENTARY NOTES

Base Calling Matlab Script

```
WTnuc='GCGGACATGGAGGACGTGCGCGGCCGCCTGGTGCAGTACCGCGGCGAGGTGCAGGCCATGCTCGGCCAGA
GCACCGAGGAGCTGCGGGTGCGCCTCGCCTCCCACCTGCGCAAGCTGCGTAAGCGGCTCCTCCGCGATGCCGATGAC
CTGCAGAAGCGCCTGGCAGTGTACCAGGCCGGGGCCCGCGAGGGCGCCGAGCGCGGCCTCAGCGCCATCCGCGAGCG
CCTGGGGCCCCTGGTGGAACAG';
%cycle through fastq files for different samples
files=dir('*.fastq');
for d=1:20
    filename=files(d).name;
    %read fastq file
    [header,seqs,qscore] = fastqread(filename);
    seqsLength = length(seqs);          % number of sequences
    seqsFile = strrep(filename,'.fastq','');      % trims off .fastq
    %create a directory with the same name as fastq file
    if exist(seqsFile,'dir');
        error('Directory already exists. Please rename or move it before
moving on.');
    end
    mkdir(seqsFile);                    % make directory
    wtLength = length(WTnuc);              % length of wildtype sequence
    %% aligning back to the wildtype nucleotide sequence
    %
    % AlN is a matrix of the nucleotide alignment
    window=1:wtLength;
    sBLength = length(seqs);         % number of sequences
    % counts number of skips
    nSkips = 0;
    ALN=repmat(' ',[sBLength wtLength]);
    % iterate through each sequencing read
    for i = 1:sBLength
        %If you only have forward read fastq files leave as is
        %If you have R1 foward and R2 is reverse fastq files uncomment the
        %next four lines of code and the subsequent end statement
%         if mod(d,2)==0;
%             reverse = seqrcomplement(seqs{i});
%             [score,alignment,start] =
swalign(reverse,WTnuc,'Alphabet','NT');
%         else

        [score,alignment,start] = swalign(seqs{i},WTnuc,'Alphabet','NT');
%         end

        % length of the sequencing read
        len = length(alignment(3,:));
        % if there is a gap in the alignment , skip = 1 and we will
        % throw away the entire read
        skip = 0;
        for j = 1:len
            if (alignment(3,j) == '-' || alignment(1,j) == '-')
                skip = 1;
                break;
            end
            %in addition if the qscore for any given base in the read is
            %below 31 the nucleotide is turned into an N (fastq qscores that
are not letters)
```

```
            if isletter(qscore{i}(start(1)+j-1))
            else
                alignment(1,j) = 'N';
            end

        end
        if skip == 0 && len>10
            ALN(i, start(2):(start(2)+length(alignment)-1))=alignment(1,:);
        end
    end
    % with the alignment matrices we can simply tally up the occurrences of
    % each nucleotide at each column in the alignment these
    % tallies ignore bases annotated as N
    % due to low qscores
    TallyNTD=zeros(5,wtLength);
    for i=1:wtLength

TallyNTD(:,i)=[sum(ALN(:,i)=='A'),sum(ALN(:,i)=='C'),sum(ALN(:,i)=='G'),sum(A
LN(:,i)=='T'),sum(ALN(:,i)=='N')];
    end
    % we then save these tally matrices in the respective folder for
    % further processing

    save(strcat(seqsFile, '/TallyNTD'), 'TallyNTD');
    dlmwrite(strcat(seqsFile, '/TallyNTD.txt'), TallyNTD, 'precision',
'%.3f', 'newline', 'pc');
end
```

INDEL Detection Matlab Script

```
WTnuc='GCGGACATGGAGGACGTGCGCGGCCGCCTGGTGCAGTACCGCGGCGAGGTGCAGGCCATGCTCGGCCAGA
GCACCGAGGAGCTGCGGGTGCGCCTCGCCTCCCACCTGCGCAAGCTGCGTAAGCGGCTCCTCCGCGATGCCGATGAC
CTGCAGAAGCGCCTGGCAGTGTACCAGGCCGGGGCCCGCGAGGGCGCCGAGCGCGGCCTCAGCGCCATCCGCGAGCG
CCTGGGGCCCCTGGTGGAACAG';
%cycle through fastq files for different samples
files=dir('*.fastq');
%specify start and width of indel window as well as length of each flank
indelstart=154;
width=30;
flank=10;

for d=1:3
    filename=files(d).name;
    %read fastq file
    [header,seqs,qscore] = fastqread(filename);
    seqsLength = length(seqs);          % number of sequences
    seqsFile = strcat(strrep(filename,'.fastq',''),'_INDELS');
    %create a directory with the same name as fastq file+_INDELS
    if exist(seqsFile,'dir');
        error('Directory already exists. Please rename or move it before
moving on.');
    end
    mkdir(seqsFile);                    % make directory
    wtLength = length(WTnuc);           % length of wildtype sequence
    sBLength = length(seqs);            % number of sequences

    % initialize counters and cell arrays
```

```
    nSkips = 0;
    notINDEL=0;
    ins={};
    dels={};
    NumIns=0;
    NumDels=0;
    % iterate through each sequencing read
    for i = 1:sBLength
    %search for 10BP sequences that should flank both sides of the "INDEL
WINDOW"
        windowstart=strfind(seqs{i},WTnuc(indelstart-flank:indelstart));

      windowend=strfind(seqs{i},WTnuc(indelstart+width:indelstart+width+flank
      ));
        %if the flanks are found proceed
        if length(windowstart)==1 && length(windowend)==1
            %if the sequence length matches the INDEL window length save as
            %not INDEL
            if windowend-windowstart==width+flank
                notINDEL=notINDEL+1;
            %if the sequence is two or more bases longer than the INDEL
            %window length save as an Insertion
            elseif windowend-windowstart>=width+flank+2
                NumIns=NumIns+1;
                ins{NumIns}=seqs{i};
            %if the sequence is two or more bases shorter than the INDEL
            %window length save as a Deletion
            elseif windowend-windowstart<=width+flank-2
                NumDels=NumDels+1;
                dels{NumDels}=seqs{i};
            %keep track of skipped sequences that are either one base
            %shorter or longer than the INDEL window width
            else
                nSkips=nSkips+1;
            end
        %keep track of skipped sequences that do not possess matching flank
        %sequences
        else
            nSkips=nSkips+1;

        end
    end

    fid=fopen(strcat(seqsFile, '/summary.txt'), 'wt');
    fprintf(fid, 'Skipped reads %i\n not INDEL %i\n Insertions %i\n Deletions
%i\n', [nSkips, notINDEL, NumIns, NumDels]);
    fclose(fid);
    save(strcat(seqsFile, '/nSkips'), 'nSkips');
    save(strcat(seqsFile, '/notINDEL'), 'notINDEL');
    save(strcat(seqsFile, '/NumIns'), 'NumIns');
    save(strcat(seqsFile, '/NumDels'), 'NumDels');
    save(strcat(seqsFile, '/dels'), 'dels');
    C = dels;
    fid = fopen(strcat(seqsFile, '/dels.txt'), 'wt');
    fprintf(fid, '"%s"\n', C{:});
    fclose(fid);
    save(strcat(seqsFile, '/ins'), 'ins');
    C = ins;
```

```
        fid = fopen(strcat(seqsFile, '/ins.txt'), 'wt');
        fprintf(fid, '"%s"\n', C{:});
        fclose(fid);

    end
```

SUPPLEMENTARY SEQUENCES

All oligonucleotides were purchased from Integrated DNA Technologies (IDT).

Primers used for generating sgRNA transfection plasmids. rev_sgRNA_plasmid was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

rev_sgRNA_plasmid	GGTGTTTCGTCCTTTCCACAAG
fwd _p53 _Y163C	GCTTGCAGATGGCCATGGCGGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_APOE4 _C158R	GAAGCGCCTGGCAGTGTACCGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_EMX1	GAGTCCGAGCAGAAGAAGAAGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_FANCF	GGAATCCCTTCTGCAGCACCGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_HEK293_2	GAACACAAAGCATAGACTGCGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_HEK293_3	GGCCCAGACTGAGCACGTGAGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_HEK293_4	GGCACTGCGGCTGGAGGTGGGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC
fwd_RNF2	GTCATCTTAGTCATTACCTGGTTTTAGAGCTAGAAATAGCAAGTTAAAATAAGGC

Sequences of all ssDNA substrates used in *in vitro* deaminase assays.

rAPOBEC1 substrate	Cy3-ATTATTATTATTCCGCGGATTTATTTATTTATTTATTTATTT
hAID/pmCDA1 substrate	Cy3-ATTATTATTATTAGCTATTTATTTATTTATTTATTTATTT
hAPOBEC3G substrate	Cy3-ATTATTATTATTCCCGGATTTATTTATTTATTTATTTATTT

Primers used for generating PCR products to serve as substrates for T7 transcription of sgRNAs for gel-based deaminase assay. rev_gRNA_T7 was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

rev_sgRNA_T7	AAAAAAAGCACCGACTCGGTG
fwd_sgRNA_T7_dsDNA_2	TAATACGACTCACTATAGGCCGCGGATTTATTTATTTAAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_3	TAATACGACTCACTATAGGTCCGCGGATTTATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_4	TAATACGACTCACTATAGGTTCCGCGGATTTATTTATTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_5	TAATACGACTCACTATAGGATTCCGCGGATTTATTTATTGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_6	TAATACGACTCACTATAGGTATTCCGCGGATTTATTTATGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_7	TAATACGACTCACTATAGGTTATTCCGCGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_8	TAATACGACTCACTATAGGATTATTCCGCGGATTTATTTGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_9	TAATACGACTCACTATAGGTATTATTCCGCGGATTTATTGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_10	TAATACGACTCACTATAGGATTATTATCCGCGGATTTATGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_11	TAATACGACTCACTATAGGTATTATATTCCGCGGATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_12	TAATACGACTCACTATAGGTTATTATATTCCGCGGATTTGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_13	TAATACGACTCACTATAGGATTATTATATTCCGCGGATTGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_14	TAATACGACTCACTATAGGTATTATTATATTCCGCGGATGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_15	TAATACGACTCACTATAGGATTATTATTATTACCGCGGAGTTTTAGAGCTAGAAATAGCA

fwd_sgRNA_T7_dsDNA_18	TAATACGACTCACTATAGGATTATTATTATTATTACCGCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_noC	TAATACGACTCACTATAGGATATTAATTTATTTATTTAAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ APOE4_C112R	TAATACGACTCACTATAGGGGAGGACGTGCGCGGCCGCCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ APOE4_C158R	TAATACGACTCACTATAGGGAAGCGCCTGGCAGTGTACCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ CTNNB1_T41A	TAATACGACTCACTATAGGCTGTGGCAGTGGCACCAGAAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ HRAS_Q61R	TAATACGACTCACTATAGGCCTCCCGGCCGGCGGTATCCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ 53_Y163C	TAATACGACTCACTATAGGGCTTGCAGATGGCCATGGCGGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ 53_Y236C	TAATACGACTCACTATAGGACACATGCAGTTGTAGTGGAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_ 53_N239D	TAATACGACTCACTATAGGTGTCACACATGTAGTTGTAGGTTTTAGAGCTAGAAATAGCA

Sequences of 80-nucleotide unlabeled strands and Cy3-labeled universal primer used in gel-based dsDNA deaminase assays.

Cy3-primer	Cy3-GTAGGTAGTTAGGATGAATGGAAGGTTGGTA
dsDNA_2	GTCCATGGATCCAGAGGTCATCCATTAAATAAATAAATCCGCGGGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_3	GTCCATGGATCCAGAGGTCATCCATAAATAAATAAATCCGCGGAAGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_4	GTCCATGGATCCAGAGGTCATCCATAATAAATAAATCCGCGGAAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_5	GTCCATGGATCCAGAGGTCATCCAAATAAATAAATCCGCGGAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_6	GTCCATGGATCCAGAGGTCATCCAATAAATAAATCCGCGGAATAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_7	GTCCATGGATCCAGAGGTCATCCATAAATAAATCCGCGGAATAAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_8	GTCCATGGATCCAGAGGTCATCCAAAATAAATCCGCGGAATAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_9	GTCCATGGATCCAGAGGTCATCCAAATAAATCCGCGGAATAATAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_10	GTCCATGGATCCAGAGGTCATCCAATAAATCCGCGGATAATAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_11	GTCCATGGATCCAGAGGTCATCCATAAATCCGCGGAATATAATAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_12	GTCCATGGATCCAGAGGTCATCCAAAATCCGCGGAATATAATAAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_13	GTCCATGGATCCAGAGGTCATCCAAATCCGCGGAATATAATAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_14	GTCCATGGATCCAGAGGTCATCCAATCCGCGGAATATAATAATAGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_15	GTCCATGGATCCAGAGGTCATCCATCCGCGGTAATAATAATAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_18	GTCCATGGATCCAGAGGTCATCCAGCGGTAATAATAATAATAATGGCTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_noC	GTCCATGGATCCAGAGGTCATCCATTAAATAAATAAATTAATATTACTATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_8U	5Cy3-GTAGGTAGTTAGGATGAATGGAAGGTTGGTGTAGATTATTATCUGCGGATTTATTGGATGACCTCTGGATCCATGGACAT
dsDNA_APOE_ C112R	GCACCTCGCCGCGGTACTGCACCAGGCGGCCGCGCACGTCCTCCATGTCTACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_APOE_ C158R	CGGCGCCCTCGCGGGCCCCGGCCTGGTACACTGCCAGGCGCTTCTGCAGTACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_CTNNB1_ T41A	GTCTTACCTGGACTCTGGAATCCATTCTGGTGCCACTGCCACAGCTCCTTACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_HRAS_ Q61R	GGAGACGTGCCTGTTGGACATCCTGGATACCGCCGGCCGGGAGGAGTACTACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_p53_ Y163C	ACCCCCGCCCGGCACCCGCGTCCGCGCCATGGCCATCTGCAAGCAGTCATACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_p53_ Y236C	AGGTTGGCTCTGACTGTACCACCATCCACTACAACTGCATGTGTAACAGTACCAACCTTCCATTCATCCTAACTACCTAC
dsDNA_p53_ N239D	TGGCTCTGACTGTACCACCATCCACTACAACTACATGTGTGACAGTTCCTACCAACCTTCCATTCATCCTAACTACCTAC

Primers used for generating PCR products to serve as substrates for T7 transcription of sgRNAs for high-throughput sequencing. rev_gRNA_T7 (above) was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

fwd_sgRNA_T7_HTS_ base	TAATACGACTCACTATAGGTTATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 1A	TAATACGACTCACTATAGGATATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 1C	TAATACGACTCACTATAGGCTATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 1G	TAATACGACTCACTATAGGGTATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 2A	TAATACGACTCACTATAGGTAATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 2C	TAATACGACTCACTATAGGTCATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 2G	TAATACGACTCACTATAGGTGATTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 3T	TAATACGACTCACTATAGGTTTTTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 3C	TAATACGACTCACTATAGGTTCTTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 3G	TAATACGACTCACTATAGGTTGTTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 4A	TAATACGACTCACTATAGGTTAATTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 4C	TAATACGACTCACTATAGGTTACTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 4G	TAATACGACTCACTATAGGTTAGTTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 5A	TAATACGACTCACTATAGGTTATATCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 5C	TAATACGACTCACTATAGGTTATCTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 5G	TAATACGACTCACTATAGGTTATGTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 6A	TAATACGACTCACTATAGGTTATTACGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 6C	TAATACGACTCACTATAGGTTATTCCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 6G	TAATACGACTCACTATAGGTTATTGCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 8A	TAATACGACTCACTATAGGTTATTTCATGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 8T	TAATACGACTCACTATAGGTTATTTCTTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 8C	TAATACGACTCACTATAGGTTATTTCCTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 9A	TAATACGACTCACTATAGGTTATTTCGAGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 9C	TAATACGACTCACTATAGGTTATTTCGCGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 9G	TAATACGACTCACTATAGGTTATTTCGGGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 10A	TAATACGACTCACTATAGGTTATTTCGTAGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 10T	TAATACGACTCACTATAGGTTATTTCGTTGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 10C	TAATACGACTCACTATAGGTTATTTCGTCGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 11A	TAATACGACTCACTATAGGTTATTTCGTGAATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 11T	TAATACGACTCACTATAGGTTATTTCGTGTATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 11C	TAATACGACTCACTATAGGTTATTTCGTGCATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 12T	TAATACGACTCACTATAGGTTATTTCGTGGTTTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 12C	TAATACGACTCACTATAGGTTATTTCGTGGCTTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 12G	TAATACGACTCACTATAGGTTATTTCGTGGGTTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 13A	TAATACGACTCACTATAGGTTATTTCGTGGAATTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ 13C	TAATACGACTCACTATAGGTTATTTCGTGGACTTATTTAGTTTTAGAGCTAGAAATAGCA

fwd_sgRNA_T7_HTS_ 13G	TAATACGACTCACTATAGGTTATTTCGTGGAGTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ multiC	TAATACGACTCACTATAGGTTCCCCCCCCGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ TCGCACCC_odd	TAATACGACTCACTATAGGCGCACCCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ CCTCGCAC_odd	TAATACGACTCACTATAGGCTCGCACGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ ACCCTCGC_odd	TAATACGACTCACTATAGGCCCTCGCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ GCACCCTC_odd	TAATACGACTCACTATAGGCACCCTCGTGGATTTATTTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ TCGCACCC_even	TAATACGACTCACTATAGGTCGCACCCGTGGATTTATTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ CCTCGCAC_even	TAATACGACTCACTATAGGCCTCGCACGTGGATTTATTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ ACCCTCGC_even	TAATACGACTCACTATAGGACCCTCGCGTGGATTTATTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ GCACCCTC_even	TAATACGACTCACTATAGGGCACCCTCGTGGATTTATTAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ EMX1	TAATACGACTCACTATAGGGAGTCCGAGCAGAAGAAGAAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ FANCF	TAATACGACTCACTATAGGGGAATCCCTTCTGCAGCACCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ HEK293_site2	TAATACGACTCACTATAGGGAACACAAAGCATAGACTGCGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ HEK293_site3	TAATACGACTCACTATAGGGGCCCAGACTGAGCACGTGAGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ HEK293_site4	TAATACGACTCACTATAGGGGCACTGCGGCTGGAGGTGGGTTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ RNF2	TAATACGACTCACTATAGGGTCATCTTAGTCATTACCTGGTTTTAGAGCTAGAAATAGCA

Sequences of *in vitro*-edited dsDNA for high-throughput sequencing (HTS). Shown are the sequences of edited strands. Reverse complements of all sequences shown were also obtained. dsDNA substrates were obtained by annealing complementary strands as described in Methods. Oligonucleotides representing the EMX1, FANCF, HEK293 site 2, HEK293 site 3, HEK293 site 4, and RNF2 loci were originally designed for use in the gel-based deaminase assay and therefore have the same 25-nt sequence on their 5'- ends (matching that of the Cy3-primer).

Base sequence	ACGTAAACGGCCACAAGTTCTTATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
1A	ACGTAAACGGCCACAAGTTCATATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
1C	ACGTAAACGGCCACAAGTTCCTATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
1G	ACGTAAACGGCCACAAGTTCGTATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
2A	ACGTAAACGGCCACAAGTTCTAATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
2C	ACGTAAACGGCCACAAGTTCTCATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
2G	ACGTAAACGGCCACAAGTTCTGATTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
3T	ACGTAAACGGCCACAAGTTCTTTTTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG

3C	ACGTAAACGGCCACAAGTTCTTCTTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
3G	ACGTAAACGGCCACAAGTTCTTGTTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
4A	ACGTAAACGGCCACAAGTTCTTAATTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
4C	ACGTAAACGGCCACAAGTTCTTACTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
4G	ACGTAAACGGCCACAAGTTCTTAGTTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
5A	ACGTAAACGGCCACAAGTTCTTATATCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
5C	ACGTAAACGGCCACAAGTTCTTATCTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
5G	ACGTAAACGGCCACAAGTTCTTATGTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
6A	ACGTAAACGGCCACAAGTTCTTATTACGTGGATTTATTTATGGCATCTTCTTCAAGGACG
6C	ACGTAAACGGCCACAAGTTCTTATTCCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
6G	ACGTAAACGGCCACAAGTTCTTATTGCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
8A	ACGTAAACGGCCACAAGTTCTTATTTCATGGATTTATTTATGGCATCTTCTTCAAGGACG
8T	ACGTAAACGGCCACAAGTTCTTATTTCTTGGATTTATTTATGGCATCTTCTTCAAGGACG
8C	ACGTAAACGGCCACAAGTTCTTATTTCCTGGATTTATTTATGGCATCTTCTTCAAGGACG
9A	ACGTAAACGGCCACAAGTTCTTATTTCGAGGATTTATTTATGGCATCTTCTTCAAGGACG
9C	ACGTAAACGGCCACAAGTTCTTATTTCGCGGATTTATTTATGGCATCTTCTTCAAGGACG
9G	ACGTAAACGGCCACAAGTTCTTATTTCGGGGATTTATTTATGGCATCTTCTTCAAGGACG
10A	ACGTAAACGGCCACAAGTTCTTATTTCGTAGATTTATTTATGGCATCTTCTTCAAGGACG
10T	ACGTAAACGGCCACAAGTTCTTATTTCGTTGATTTATTTATGGCATCTTCTTCAAGGACG
10C	ACGTAAACGGCCACAAGTTCTTATTTCGTCGATTTATTTATGGCATCTTCTTCAAGGACG
11A	ACGTAAACGGCCACAAGTTCTTATTTCGTGAATTTATTTATGGCATCTTCTTCAAGGACG
11T	ACGTAAACGGCCACAAGTTCTTATTTCGTGTATTTATTTATGGCATCTTCTTCAAGGACG
11C	ACGTAAACGGCCACAAGTTCTTATTTCGTGCATTTATTTATGGCATCTTCTTCAAGGACG
12T	ACGTAAACGGCCACAAGTTCTTATTTCGTGGTTTTATTTATGGCATCTTCTTCAAGGACG
12C	ACGTAAACGGCCACAAGTTCTTATTTCGTGGCTTTATTTATGGCATCTTCTTCAAGGACG
12G	ACGTAAACGGCCACAAGTTCTTATTTCGTGGGTTTATTTATGGCATCTTCTTCAAGGACG
13A	ACGTAAACGGCCACAAGTTCTTATTTCGTGGAATTATTTATGGCATCTTCTTCAAGGACG
13C	ACGTAAACGGCCACAAGTTCTTATTTCGTGGACTTATTTATGGCATCTTCTTCAAGGACG
13G	ACGTAAACGGCCACAAGTTCTTATTTCGTGGAGTTATTTATGGCATCTTCTTCAAGGACG
multiC	ACGTAAACGGCCACAAGTTCTTCCCCCCCCGATTTATTTATGGCATCTTCTTCAAGGACG
TCGCACCC_odd	ACGTAAACGGCCACAAGTTTCGCACCCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
CCTCGCAC_odd	ACGTAAACGGCCACAAGTTCCTCGCACGTGGATTTATTTATGGCATCTTCTTCAAGGACG
ACCCTCGC_odd	ACGTAAACGGCCACAAGTTACCCTCGCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
GCACCCTC_odd	ACGTAAACGGCCACAAGTTGCACCCTCGTGGATTTATTTATGGCATCTTCTTCAAGGACG
TCGCACCC_even	ACGTAAACGGCCACAAGTATTCGCACCCGTGGATTTATTATGGCATCTTCTTCAAGGACG
CCTCGCAC_even	ACGTAAACGGCCACAAGTATCCTCGCACGTGGATTTATTATGGCATCTTCTTCAAGGACG
ACCCTCGC_even	ACGTAAACGGCCACAAGTATACCCTCGCGTGGATTTATTATGGCATCTTCTTCAAGGACG
GCACCCTC_even	ACGTAAACGGCCACAAGTATGCACCCTCGTGGATTTATTATGGCATCTTCTTCAAGGACG

EMX1_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTAGGCCTGAGTCCGAGCAGAAGAAGAAGGGCTCCCATCACATCAACCGGTG
FANCF_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACTCATGGAATCCCTTCTGCAGCACCTGGATCGCTTTTCCGAGCTTCTGG
HEK293_site2_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTAAACTGGAACACAAAGCATAGACTGCGGGGCGGGCCAGCCTGAATAGCTG
HEK293_site3_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACTTGGGGCCCAGACTGAGCACGTGATGGCAGAGGAAAGGAAGCCCTGCT
HEK293_site4_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACCGGTGGCACTGCGGCTGGAGGTGGGGGTTAAAGCGGAGACTCTGGTGC
RNF2_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTATGGCAGTCATCTTAGTCATTACCTGAGGTGTTCGTTGTAACTCATATAA

Primers for HTS of *in vitro* edited dsDNA.

fwd_invitro_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNACGTAAACGGCCACAA
rev_invitro_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCGTCCTTGAAGAAGATGC
fwd_invitro_HEK_targets	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGTAGGTAGTTAGGATGAATGGAA
rev_EMX1_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTCACCGGTTGATGTGATGG
rev_FANCF_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTCCAGAAGCTCGGAAAAGC
rev_HEK293_site2_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTCAGCTATTCAGGCTGGC
rev_HEK293_site3_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTAGCAGGGCTTCCTTTC
rev_HEK293_site4_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCACCAGAGTCTCCG
rev_RNF2_invitro	TGGAGTTCAGACGTGTGCTCTTCCGATCTTTATATGAGTTACAACGAACACC

Primers for HTS of on-target and off-target sites from all mammalian cell culture experiments.

fwd_EMX1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCAGCTCAGCCTGAGTGTTGA
rev_EMX1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTCGTGGGTTTGTGGTTGC
fwd_FANCF_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCATTGCAGAGAGGCGTATCA
rev_FANCF_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGGGGTCCCAGGTGCTGAC
fwd_HEK293_site2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCAGCCCCATCTGTCAAACT
rev_HEK293_site2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTGAATGGATTCCTTGGAAACAATGA
fwd_HEK293_site3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNATGTGGGCTGCCTAGAAAGG
rev_HEK293_site3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCCCAGCCAAACTTGTCAACC
fwd_HEK293_site4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGAACCCAGGTAGCCAGAGAC
rev_HEK293_site4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTCCTTTCAACCCGAACGGAG
fwd_RNF2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNACGTCTCATATGCCCCTTGG
rev_RNF2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTACGTAGGAATTTTGGTGGGACA
fwd_p53_Y163C_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTGCCCTGACTTTCAACTCTGT
rev_p53_Y163C_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCAACCAGCCCTGTCGTCTC
fwd_APOE4_C158R_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGACATGGAGGACGTGCG
rev_APOE4_C158R_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTCCTGTAGCGGCTGGCC
fwd_EMX1_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTGCCCAATCATTGATGCTTTT
rev_EMX1_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTAGAAACATTTACCATAGACTATCACCT

fwd_EMX1_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAGTAGCCTCTTTCTCAATGTGC
rev_EMX1_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCTTTCACAAGGATGCAGTCT
fwd_EMX1_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGAGCTAGACTCCGAGGGGA
rev_EMX1_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTCCTCGTCCTGCTCTCACTT
fwd_EMX1_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAGAGGCTGAAGAGGAAGACCA
rev_EMX1_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGGCCCAGCTGTGCATTCTAT
fwd_EMX1_off6_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCAAGAGGGCCAAGTCCTG
rev_EMX1_off6_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCAGCGAGGAGTGACAGCC
fwd_EMX1_off7_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCACTCCACCTGATCTCGGGG
rev_EMX1_off7_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCGAGGAGGGAGGGAGCAG
fwd_EMX1_off8_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNACCACAAATGCCCAAGAGAC
rev_EMX1_off8_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGACACAGTCAAGGGCCGG
fwd_EMX1_off9_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCCACCTTTGAGGAGGCAAA
rev_EMX1_off9_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTTCCATCTGAGAAGAGAGTGGT
fwd_EMX1_off10_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGTCATACCTTGGCCCTTCCT
rev_EMX1_off10_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTCCCTAGGCCCACACCAG
fwd_FANCF_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAACCCACTGAAGAAGCAGGG
rev_FANCF_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGGTGCTTAATCCGGCTCCAT
fwd_FANCF_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTCCAGTGTTTCCATCCCGAA
rev_FANCF_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCCTCTGACCTCCACAACTCT
fwd_FANCF_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCTGGGTACAGTTCTGCGTGT
rev_FANCF_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTCACTCTGAGCATCGCCAAG
fwd_FANCF_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGTTTAGAGCCAGTGAACTAGAG
rev_FANCF_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCAAGACAAAATCCTCTTTATACTTTG
fwd_FANCF_off5_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGGAGGGGACGGCCTTAC
rev_FANCF_off5_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCCTCTGGCGAACATGGC
fwd_FANCF_off6_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTCCTGGTTAAGAGCATGGGC
rev_FANCF_off6_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGATTGAGTCCCCACAGCACA
fwd_FANCF_off7_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCAGTGTTTCCCATCCCCAA
rev_FANCF_off7_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTGACCTCCACAACTGGAAAAT
fwd_FANCF_off8_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGCTTCCAGACCCACCTGAAG
rev_FANCF_off8_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTACCGAGGAAAATTGCTTGTCG
fwd_HEK293_site2_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGTGTGGAGAGTGAGTAAGCCA
rev_HEK293_site2_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTACGGTAGGATGATTTCAGGCA
fwd_HEK293_site2_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCACAAAGCAGTGTAGCTCAGG
rev_HEK293_site2_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTTTTTTGGTACTCGAGTGTTATTCAG
fwd_HEK2_ChIP_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGACAGGCTCAGGAAAGCTGT
rev_HEK2_ChIP_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTACACAAGCCTTTCTCCAGGG
fwd_HEK2_ChIP_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAATAGGGGGTGAGACTGGGG

rev_HEK2_ChIP_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCCTCAGACGAGACTTGAGG
fwd_HEK2_ChIP_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGCCAGCAGGAAAGGAATCT
rev_HEK2_ChIP_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTGACTGCACCTGTAGCCATG
fwd_HEK2_ChIP_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTCAAGGAAATCACCCTGCCC
rev_HEK2_ChIP_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTAACTTCCTTGGTGTGCAGCT
fwd_HEK2_ChIP_off5_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNATGGGCTCAGCTACGTCATG
rev_HEK2_ChIP_off5_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTAATAGCAGTGTGGTGGGCAA
fwd_HEK293_site3_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTCCCCTGTTGACCTGGAGAA
rev_HEK293_site3_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCACTGTACTTGCCCTGACCA
fwd_HEK293_site3_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTTGGTGTTGACAGGGAGCAA
rev_HEK293_site3_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTGAGATGTGGGCAGAAGGG
fwd_HEK293_site3_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTGAGAGGGAACAGAAGGGCT
rev_HEK293_site3_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGTCCAAAGGCCCAAGAACCT
fwd_HEK293_site3_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTCCTAGCACTTTGGAAGGTCG
rev_HEK293_site3_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCTCATCTTAATCTGCTCAGCC
fwd_HEK293_site3_off5_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAAAGGAGCAGCTCTTCCTGG
rev_HEK293_site3_off5_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGTCTGCACCATCTCCCACAA
fwd_HEK3_ChIP_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCGCACATCCCTTGTCTCTCT
rev_HEK3_ChIP_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTACTGGAGCACACCCCAAG
fwd_HEK3_ChIP_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTGGGTCACGTAGCTTTGGTC
rev_HEK3_ChIP_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTGGTGGCCATGTGCAACTAA
fwd_HEK3_ChIP_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCTACTACGTGCCAGCTCAGG
rev_HEK3_ChIP_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTACCTCCCCTCCTCACTAACC
fwd_HEK3_ChIP_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGCCTCAGCTCCATTTCCTGT
rev_HEK3_ChIP_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTAACCTTTATGGCACCAGGGG
fwd_HEK3_ChIP_off5_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGAGCTCAGCATTAGCAGGCT
rev_HEK3_ChIP_off5_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTTCCTGGCTTTCCGATTCCC
fwd_HEK293_site4_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGCATGGCTTCTGAGACTCA
rev_HEK293_site4_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGTCTCCCTTGCACTCCCTGTCTTT
fwd_HEK293_site4_off2_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTTTGGCAATGGAGGCATTGG
rev_HEK293_site4_off2_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGAAGAGGCTGCCCATGAGAG
fwd_HEK293_site4_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGGTCTGAGGCTCGAATCCTG
rev_HEK293_site4_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTGTGGCCTCCATATCCCTG
fwd_HEK293_site4_off4_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNTTTCCACCAGAACTCAGCCC
rev_HEK293_site4_off4_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCCTCGGTTCCTCCACAACAC
fwd_HEK293_site4_off5_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCACGGGAAGGACAGGAGAAG
rev_HEK293_site4_off5_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCAGGGGAGGGATAAAGCAG
fwd_HEK293_site4_off6_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCACGGGAGATGGCTTATGT
rev_HEK293_site4_off6_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCACATCCTCACTGTGCCACT

fwd_HEK293_site4_off7_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGTCAGTCTCGGCCCCTCA
rev_HEK293_site4_off7_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGCCACTGTAAAGCTCTTGGG
fwd_HEK293_site4_off8_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNAGGGTAGAGGGACAGAGCTG
rev_HEK293_site4_off8_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTGGACCCCACATAGTCAGTGC
fwd_HEK293_site4_off9_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGCTGTCAGCCCTATCTCCATC
rev_HEK293_site4_off9_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTTGGGCAATTAGGACAGGGAC
fwd_HEK293_site4_off10_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGCAGCGGAGGAGGTAGATTG
rev_HEK293_site4_off10_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCTCAGTACCTGGAGTCCCGA
fwd_HEK4_ChIP_off1_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNGTGCAATTGGAGGAGGAGCT
rev_HEK4_ChIP_off1_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCACCAGCTACAGGCAGAACA
fwd_HEK4_ChIP_off3_HTS	ACACTCTTTCCCTACACGACGCTCTTCCGATCTNNNNCCTACCCCCAACACAGATGG
rev_HEK4_ChIP_off3_HTS	TGGAGTTCAGACGTGTGCTCTTCCGATCTCCACACAACTCAGGTCCTCC

Sequences of single-stranded oligonucleotide donor templates (ssODNs) used in HDR studies.

EMX1 sense
TCATCTGTGCCCCTCCCTCCCTGGCCCAGGTGAAGGTGTGGTTCCAGAACCGGAGGACAAAGTACA
AACGGCAGAAGCTGGAGGAGGAAGGGCCTGAGTTTGAGCAGAAGAAGAAGGGCTCCCATCACATC
AACCGGTGGCGCATTGCCACGAAGCAGGCCAATGGGGAGGACATCGATGTCACCTCCAATGACTAG
GGT

EMX1 antisense
ACCCTAGTCATTGGAGGTGACATCGATGTCCTCCCCATTGGCCTGCTTCGTGGCAATGCGCCACCG
GTTGATGTGATGGGAGCCCTTCTTCTTCTGCTCAAACTCAGGCCCTTCCTCCTCCAGCTTCTGCCGT
TTGTACTTTGTCCTCCGGTTCTGGAACCACACCTTCACCTGGGCCAGGGAGGGAGGGGCACAGATG
A

HEK293 site 3 sense
CATGCAATTAGTCTATTTCTGCTGCAAGTAAGCATGCATTTGTAGGCTTGATGCTTTTTTTCTGCTTCT
CCAGCCCTGGCCTGGGTCAATCCTTGGGGCTTAGACTGAGCACGTGATGGCAGAGGAAAGGAAGC
CCTGCTTCCTCCAGAGGGCGTCGCAGGACAGCTTTTCCTAGACAGGGGCTAGTATGTGCAGCTCCT

HEK293 site 3 antisense
AGGAGCTGCACATACTAGCCCCTGTCTAGGAAAAGCTGTCCTGCGACGCCCTCTGGAGGAAGCAGG
GCTTCCTTTCCTCTGCCATCACGTGCTCAGTCTAAGCCCCAAGGATTGACCCAGGCCAGGGCTGGA
GAAGCAGAAAAAAAGCATCAAGCCTACAAATGCATGCTTACTTGCAGCAGAAATAGACTAATTGCATG

HEK293 site 4 sense
GGCTGACAAAGGCCGGGCTGGGTGGAAGGAAGGGAGGAAGGGCGAGGCAGAGGGTCCAAAGCAG
GATGACAGGCAGGGGCACCGCGGCGCCCCGGTGGCATTGCGGCTGGAGGTGGGGGTTAAAGCGG
AGACTCTGGTGCTGTGTGACTACAGTGGGGGCCCTGCCCTCTCTGAGCCCCCGCCTCCAGGCCTGT
GTGTGT

HEK293 site 4 antisense
ACACACACAGGCCTGGAGGCGGGGGCTCAGAGAGGGCAGGGCCCCCACTGTAGTCACACAGCACC
AGAGTCTCCGCTTTAACCCCCACCTCCAGCCGCAATGCCACCGGGGCGCCGCGGTGCCCCTGCCT
GTCATCCTGCTTTGGACCCTCTGCCTCGCCCTTCCTCCCTTCCTTCCACCCAGCCCGGCCTTTGTCA
GCC

APOE4 sense
AGCACCGAGGAGCTGCGGGTGCGCCTCGCCTCCCACCTGCGCAAGCTGCGTAAGCGGCTCCTCCG
CGATGCCGATGACCTGCAGAAGTGCCTGGCAGTGTACCAGGCCGGGGCCCGCGAGGGCGCCGAG

CGCGGCCTCAGCGCCATCCGCGAGCGCCTGGGGCCCCTGGTGGAACAGGGCCGCGTGCGGGCCG
CCACTGT

APOE4 antisense
ACAGTGGCGGCCCGCACGCGGCCCTGTTCCACCAGGGGCCCCAGGCGCTCGCGGATGGCGCTGA
GGCCGCGCTCGGCGCCCTCGCGGGCCCCGGCCTGGTACACTGCCAGGCACTTCTGCAGGTCATCG
GCATCGCGGAGGAGCCGCTTACGCAGCTTGCGCAGGTGGGAGGCGAGGCGCACCCGCAGCTCCT
CGGTGCT

p53 Y163C sense
ACTCCCCTGCCCTCAACAAGATGTTTTGCCAACTGGCCAAGACCTGCCCTGTGCAGCTGTGGGTTGA
TTCCACACCCCCGCCCGGCACCCGCGTCCGCGCCATGGCCATCTACAAGCAGTCACAGCACATGAC
GGAGGTTGTGAGGCGCTGCCCCCACCATGAGCGCTGCTCAGATAGCGATGGTGAGCAGCTGGGGC
TG

p53 Y163C antisense
CAGCCCCAGCTGCTCACCATCGCTATCTGAGCAGCGCTCATGGTGGGGGCAGCGCCTCACAACCTC
CGTCATGTGCTGTGACTGCTTGTAGATGGCCATGGCGCGGACGCGGGTGCCGGGCGGGGGTGTGG
AATCAACCCACAGCTGCACAGGGCAGGTCTTGGCCAGTTGGCAAAACATCTTGTTGAGGGCAGGGG
AGT

Deaminase gene gBlocks gene fragments.

hAID
CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGGATAGCCTCTTGATGAATAGACGCAAGTTCCT
GTATCAGTTTAAAAACGTGAGATGGGCAAAAGGCCGACGAGAGACATATCTGTGCTATGTCGTTAAG
CGCAGAGATTCAGCCACCAGTTTCTCTCTCGACTTCGGCTACCTGCGGAACAAGAATGGTTGCCATG
TTGAGCTCCTGTTCCTGAGGTATATCAGCGACTGGGATTTGGACCCAGGGCGGTGCTATAGGGTGA
CATGGTTTACCTCCTGGTCACCTTGTTATGACTGCGCGCGGCATGTTGCCGATTTTCTGAGAGGGAA
CCCTAACCTGTCTCTGAGGATCTTCACCGCGCGACTGTACTTCTGTGAGGACCGGAAAGCCGAACC
CGAGGGACTGAGACGCCTCCACAGAGCGGGTGTGCAGATTGCCATAATGACCTTTAAGGACTACTT
CTACTGCTGGAACACCTTCGTCGAAAATCACGAGCGGACTTTCAAGGCTTGGGAAGGATTGCATGAA
AACAGCGTCAGGCTTTCCAGGCAGCTTCGCCGCATTCTTCTCCCGTTGTACGAGGTTGATGACCTCA
GAGATGCCTTTAGAACACTGGGACTGTAGGCGGCCGCTCGATTGGTTTGGTGTGGCTCTAA

rAPOBEC1 (mammalian)
CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGAGCTCAGAGACTGGCCCAGTGGCTGTGGACC
CCACATTGAGACGGCGGATCGAGCCCCATGAGTTTGAGGTATTCTTCGATCCGAGAGAGCTCCGCA
AGGAGACCTGCCTGCTTTACGAAATTAATTGGGGGGGCCGGCACTCCATTTGGCGACATACATCACA
GAACACTAACAAGCACGTCGAAGTCAACTTCATCGAGAAGTTCACGACAGAAAGATATTTCTGTCCG
AACACAAGGTGCAGCATTACCTGGTTTCTCAGCTGGAGCCCATGCGGCGAATGTAGTAGGGCCATC
ACTGAATTCCTGTCAAGGTATCCCCACGTCACTCTGTTTATTTACATCGCAAGGCTGTACCACCACGC
TGACCCCCGCAATCGACAAGGCCTGCGGGATTTGATCTCTTCAGGTGTGACTATCCAAATTATGACT
GAGCAGGAGTCAGGATACTGCTGGAGAAACTTTGTGAATTATAGCCCGAGTAATGAAGCCCACTGG
CCTAGGTATCCCCATCTGTGGGTACGACTGTACGTTCTTGAACTGTACTGCATCATACTGGGCCTGC
CTCCTTGTCTCAACATTCTGAGAAGGAAGCAGCCACAGCTGACATTCTTTACCATCGCTCTTCAGTCT
TGTCATTACCAGCGACTGCCCCCACACATTCTCTGGGCCACCGGGTTGAAATGAGCGGCCGCTCGA
TTGGTTTGGTGTGGCTCTAA

pmCDA1
CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGACAGACGCTGAATATGTTAGGATCCATGAAAA
ACTGGATATCTATACATTTAAGAAGCAGTTCTTCAATAACAAAAAGTCAGTATCTCACAGATGCTATGT
CCTGTTCGAACTCAAGAGAAGAGGAGAAAGGCGGGCCTGTTTCTGGGGGTACGCGGTTAATAAACC
CCAGTCCGGGACCGAGAGGGGGATTCACGCCGAGATCTTTTCAATTAGGAAGGTTGAAGAGTATCT
TCGCGACAATCCCGGTCAGTTCACAATTAACTGGTACAGCTCCTGGAGCCCTTGCGCTGATTGCGCC

GAGAAAATACTCGAATGGTACAACCAGGAGTTGAGAGGCAATGGCCACACTCTCAAGATTTGGGCTT
GCAAGCTTTACTACGAGAAGAACGCGAGAAATCAGATTGGCTTGTGGAACCTCAGGGACAACGGGG
TCGGGTTGAATGTTATGGTGTCCGAACATTACCAGTGCTGTAGAAAGATCTTCATTCAGTCCAGTCAC
AATCAGCTGAACGAGAACAGATGGCTGGAGAAAACACTGAAACGGGCAGAGAAAAGGCGCTCAGAG
CTGAGTATCATGATCCAGGTCAAAATCCTGCATACAACCAAAAGCCCGGCTGTATAAGCGGCCGCTC
GATTGGTTTGGTGTGGCTCTAA

hAPOBEC3G
CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGGAGCTGAAGTATCACCCTGAGATGCGGTTTTT
CCACTGGTTTAGTAAGTGGCGCAAACTTCATCGGGATCAGGAGTATGAAGTGACCTGGTATATCTCT
TGGTCTCCCTGCACAAAATGTACACGCGACATGGCCACATTTCTGGCCGAGGATCCAAAGGTGACG
CTCACAATCTTTGTGGCCCGCCTGTATTATTTCTGGGACCCGGATTATCAGGAGGCACTTAGGTCAT
TGTGCCAAAAGCGCGACGGACCACGGGCGACTATGAAAATCATGAATTATGACGAATTCCAGCATTG
CTGGAGTAAGTTTGTGTACAGCCAGCGGGAGCTGTTCGAGCCCTGGAACAATCTTCCCAAGTACTAC
ATACTGCTTCACATTATGTTGGGGGAGATCCTTCGGCACTCTATGGATCCTCCTACCTTTACGTTTAA
CTTTAATAATGAGCCTTGGGTTCGCGGGCGCCATGAAACCTATTTGTGCTACGAGGTCGAGCGGATG
CATAATGATACGTGGGTCCTGCTGAATCAGAGGAGGGGGTTTCTGTGTAACCAGGCTCCACATAAAC
ATGGATTTCTCGAGGGGCGGCACGCCGAACTGTGTTTCCTTGATGTGATACCTTTCTGGAAGCTCGA
CCTTGATCAAGATTACAGGGTGACGTGTTTCACCTCCTGGTCACCCTGCTTCAGTTGCGCCCAAGAG
ATGGCTAAATTTATCAGTAAGAACAAGCATGTGTCCCTCTGTATTTTTACAGCCAGAATTTATGATGAC
CAGGGCCGGTGCCAGGAGGGGCTGCGGACACTCGCTGAGGCGGGCGCGAAGATCAGCATAATGA
CATACTCCGAATTCAAACACTGTTGGGACACTTTTGTGGACCACCAGGGCTGCCCATTTCAGCCGTG
GGATGGGCTCGACGAACATAGTCAGGATCTCTCAGGCCGGCTGCGAGCCATATTGCAGAACCAGGA
GAATTAGGCGGCCGCTCGATTGGTTTGGTGTGGCTCTAA

rAPOBEC1(*E. Coli*)
GGCCGGGGATTCTAGAAATAATTTTGTTTAACTTTAAGAAGGAGATATACCATGGATGTCTTCTGAAA
CCGGTCCGGTTGCGGTTGACCCGACCCTGCGTCGTCGTATCGAACCGCACGAATTCGAAGTTTTCT
TCGACCCGCGTGAACTGCGTAAAGAAACCTGCCTGCTGTACGAAATCAACTGGGGTGGTCGTCACT
CTATCTGGCGTCACACCTCTCAGAACACCAACAAACACGTTGAAGTTAACTTCATCGAAAAATTCACC
ACCGAACGTTACTTCTGCCCGAACACCCGTTGCTCTATCACCTGGTTCCTGTCTTGGTCTCCGTGCG
GTGAATGCTCTCGTGCGATCACCGAATTCCTGTCTCGTTACCCGCACGTTACCCTGTTCATCTACATC
GCGCGTCTGTACCACCACGCGGACCCGCGTAACCGTCAGGGTCTGCGTGACCTGATCTCTTCTGGT
GTTACCATCCAGATCATGACCGAACAGGAATCTGGTTACTGCTGGCGTAACTTCGTTAACTACTCTCC
GTCTAACGAAGCGCACTGGCCGCGTTACCCGCACCTGTGGGTTCGTCTGTACGTTCTGGAACTGTA
CTGCATCATCCTGGGTCTGCCGCCGTGCCTGAACATCCTGCGTCGTAAACAGCCGCAGCTGACCTT
CTTCACCATCGCGCTGCAGTCTTGCCACTACCAGCGTCTGCCGCCGCACATCCTGTGGGCGACCGG
TCTGAAAGGTGGTAGTGGAGGGAGCGGCGGTTCAATGGATAAGAAATAC

Amino Acid Sequences of BE1, BE2, and BE3.

BE1 for *E. Coli* expression (His_6-rAPOBEC1-XTEN-dCas9)
MGSSHHHHHHMSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTN
KHVEVNFIEKFTTERYFCPNTRCSITWFLSWSPCGECSRAITEFLSRYPHVTLFIYIARLYHHADPRNRQGL
RDLISSGVTIQIMTEQESGYCWRNFVNYSPSNEAHWPRYPHLWVRLYVLELYCIILGLPPCLNILRRKQPQ
LTFFTIALQSCHYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSK
KFKVLGNTDRHSIKKNLIGALLFDSGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEMAKVDDSFFHRL
EESFLVEEDKKHERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKADLRLIYLALAHMIKFRGHFLIEGDL
NPDNSDVDKLFIQLVQTYNQLFEENPINASGVDAKAILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSL
GLTPNFKSNFDLAEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLAAKNLSDAILLSDILRVNTEITKAPLSA
SMIKRYDEHHQDLTLLKALVRQQLPEKYKEIFFDQSKNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELL
VKLNREDLLRKQRTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRF
AWMTRKSEETITPWNFEEVVDKGASAQSFIERMTNFDKNLPNEKVLPKHSLLYEYFTVYNELTKVKYVTE
GMRKPAFLSGEQKKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFNASLGTYHDLLKIIKDK
DFLDNEENEDILEDIVLTLTLFEDREMIEERLKTYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDKQS
GKTILDFLKSDGFANRNFMQLIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPAIKKGILQTVKVVDEL

VKVMGRHKPENIVIEMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYYLQ
NGRDMYVDQELDINRLSDYDVDAIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQL
LNAKLITQRKFDNLTKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLK
SKLVSDFRKDFQFYKVREINNYHHAHDAYLNAVVGTALIKKYPKLESEFVYGDYKVYDVRKMIAKSEQEIG
KATAKYFFYSNIMNFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVLSMPQVNIVKKTEVQT
GGFSKESILPKRNSDKLIARKKDWDPKKYGGFDSPTVAYSVLVVAKVEKGKSKKLKSVKELLGITIMERSS
FEKNPIDFLEAKGYKEVKKDLIIKLPKYSLFELENGRKRMLASAGELQKGNELALPSKYVNFLYLASHYEKL
KGSPEDNEQKQLFVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNL
GAPAAFKYFDTTIDRKRYTSTKEVLDATLIHQSITGLYETRIDLSQLGGDSGGSPKKKRKV

BE1 for Mammalian expression (rAPOBEC1-XTEN-dCas9-NLS)
MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNFIEKF
TTERYFCPNTRCSITWFLSWSPCGECSRAITEFLSRYPHVTLFIYIARLYHHADPRNRQGLRDLISSGVTIQ
IMTEQESGYCWRNFVNYSPSNEAHWPRYPHLWVRLYVLELYCIILGLPPCLNILRRKQPQLTFFTIALQSC
HYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSKKFKVLGNTDR
HSIKKNLIGALLFDSGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEMAKVDDSFFHRLEESFLVEEDKK
HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKADLRLIYLALAHMIKFRGHFLIEGDLNPDNSDVDKLF
IQLVQTYNQLFEENPINASGVDAKAILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL
AEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLAAKNLSDAILLSDILRVNTEITKAPLSASMIKRYDEHHQ
DLTLLKALVRQQLPEKYKEIFFDQSKNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELLVKLNREDLLRKQ
RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAWMTRKSEETITP
WNFEEVVDKGASAQSFIERMTNFDKNLPNEKVLPKHSLLYEYFTVYNELTKVKYVTEGMRKPAFLSGEQ
KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFNASLGTYHDLLKIIKDKDFLDNEENEDILE
DIVLTLTLFEDREMIEERLKTYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDKQSGKTILDFLKSDGFA
NRNFMQLIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPAIKKGILQTVKVVDELVKVMGRHKPENIVI
EMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYYLQNGRDMYVDQELDI
NRLSDYDVDAIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQLLNAKLITQRKFDNL
TKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKSKLVSDFRKDFQF
YKVREINNYHHAHDAYLNAVVGTALIKKYPKLESEFVYGDYKVYDVRKMIAKSEQEIGKATAKYFFYSNIM
NFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVLSMPQVNIVKKTEVQTGGFSKESILPKRN
SDKLIARKKDWDPKKYGGFDSPTVAYSVLVVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKG
YKEVKKDLIIKLPKYSLFELENGRKRMLASAGELQKGNELALPSKYVNFLYLASHYEKLKGSPEDNEQKQL
FVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI
DRKRYTSTKEVLDATLIHQSITGLYETRIDLSQLGGDSGGSPKKKRKV

BE2 (rAPOBEC1-XTEN-dCas9-UGI-NLS)
MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNFIEKF
TTERYFCPNTRCSITWFLSWSPCGECSRAITEFLSRYPHVTLFIYIARLYHHADPRNRQGLRDLISSGVTIQ
IMTEQESGYCWRNFVNYSPSNEAHWPRYPHLWVRLYVLELYCIILGLPPCLNILRRKQPQLTFFTIALQSC
HYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSKKFKVLGNTDR
HSIKKNLIGALLFDSGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEMAKVDDSFFHRLEESFLVEEDKK
HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKADLRLIYLALAHMIKFRGHFLIEGDLNPDNSDVDKLF
IQLVQTYNQLFEENPINASGVDAKAILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL
AEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLAAKNLSDAILLSDILRVNTEITKAPLSASMIKRYDEHHQ
DLTLLKALVRQQLPEKYKEIFFDQSKNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELLVKLNREDLLRKQ
RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAWMTRKSEETITP
WNFEEVVDKGASAQSFIERMTNFDKNLPNEKVLPKHSLLYEYFTVYNELTKVKYVTEGMRKPAFLSGEQ
KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFNASLGTYHDLLKIIKDKDFLDNEENEDILE
DIVLTLTLFEDREMIEERLKTYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDKQSGKTILDFLKSDGFA
NRNFMQLIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPAIKKGILQTVKVVDELVKVMGRHKPENIVI
EMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYYLQNGRDMYVDQELDI
NRLSDYDVDAIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQLLNAKLITQRKFDNL
TKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKSKLVSDFRKDFQF
YKVREINNYHHAHDAYLNAVVGTALIKKYPKLESEFVYGDYKVYDVRKMIAKSEQEIGKATAKYFFYSNIM
NFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVLSMPQVNIVKKTEVQTGGFSKESILPKRN
SDKLIARKKDWDPKKYGGFDSPTVAYSVLVVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKG

YKEVKKDLIIKLPKYSLFELENGRKRMLASAGELQKGNELALPSKYVNFLYLASHYEKLKGSPEDNEQKQL
FVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI
DRKRYTSTKEVLDATLIHQSITGLYETRIDLSQLGGDSGGSTNLSDIIEKETGKQLVIQESILMLPEEVEEVIG
NKPESDILVHTAYDESTDENVMLLTSDAPEYKPWALVIQDSNGENKIKMLSGGSPKKKRKV

BE3 (rAPOBEC1-XTEN-Cas9n-UGI-NLS)
MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNFIEKF
TTERYFCPNTRCSITWFLSWSPCGECSRAITEFLSRYPHVTLFIYIARLYHHADPRNRQGLRDLISSGVTIQ
IMTEQESGYCWRNFVNYSPSNEAHWPRYPHLWVRLYVLELYCIILGLPPCLNILRRKQPQLTFFTIALQSC
HYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSKKFKVLGNTDR
HSIKKNLIGALLFDSGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEMAKVDDSFFHRLEESFLVEEDKK
HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKADLRLIYLALAHMIKFRGHFLIEGDLNPDNSDVDKLF
IQLVQTYNQLFEENPINASGVDAKAILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL
AEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLAAKNLSDAILLSDILRVNTEITKAPLSASMIKRYDEHHQ
DLTLLKALVRQQLPEKYKEIFFDQSKNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELLVKLNREDLLRKQ
RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAWMTRKSEETITP
WNFEEVVDKGASAQSFIERMTNFDKNLPNEKVLPKHSLLYEYFTVYNELTKVKYVTEGMRKPAFLSGEQ
KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFNASLGTYHDLLKIIKDKDFLDNEENEDILE
DIVLTLTLFEDREMIEERLKTYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDKQSGKTILDFLKSDGFA
NRNFMQLIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPAIKKGILQTVKVVDELVKVMGRHKPENIVI
EMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYYLQNGRDMYVDQELDI
NRLSDYDVDHIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQLLNAKLITQRKFDNL
TKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKSKLVSDFRKDFQF
YKVREINNYHHAHDAYLNAVVGTALIKKYPKLESEFVYGDYKVYDVRKMIAKSEQEIGKATAKYFFYSNIM
NFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVLSMPQVNIVKKTEVQTGGFSKESILPKRN
SDKLIARKKDWDPKKYGGFDSPTVAYSVLVVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKG
YKEVKKDLIIKLPKYSLFELENGRKRMLASAGELQKGNELALPSKYVNFLYLASHYEKLKGSPEDNEQKQL
FVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI
DRKRYTSTKEVLDATLIHQSITGLYETRIDLSQLGGDSGGSTNLSDIIEKETGKQLVIQESILMLPEEVEEVIG
NKPESDILVHTAYDESTDENVMLLTSDAPEYKPWALVIQDSNGENKIKMLSGGSPKKKRKV

SUPPLEMENTARY TABLES

Supplementary Table 1. Activities of BE1, BE2, and BE3 at EMX1 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the EMX1 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus the top ten known Cas9 off-target loci for the EMX1 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 19). EMX1 off-target 5 locus did not amplify and is not shown. Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers for each sequence.

																							GUIDE-seq counts
EMX1 on target	G	A	G	T	C	C	G	A	G	C	A	G	A	A	G	A	A	G	A	A	G	G	G	4521
untreated					0.1	0.0				0.0														
BE1					4.2	4.4				0.0														
BE2					9.1	9.3				0.0														
BE3					26.0	26.6				0.0														
EMX1 off target 1	G	A	G	T	T	A	G	A	G	C	A	G	A	A	G	A	A	G	A	A	A	G	G	3123
untreated										0.0														
BE1										0.0														
BE2										0.0														
BE3										0.0														
EMX1 off target 2	G	A	G	T	C	T	A	A	G	C	A	G	A	A	G	A	A	G	A	A	G	A	G	1445
untreated					0.0					0.0														
BE1					1.0					0.0														
BE2					3.6					0.0														
BE3					8.5					0.0														
EMX1 off target 3	G	A	G	G	C	C	G	A	G	C	A	G	A	A	G	A	A	A	G	A	C	G	G	700
untreated					0.0	0.0				0.0											0.0			
BE1					0.0	0.0				0.0											0.0			
BE2					0.8	0.9				0.0											0.0			
BE3					5.1	5.2				0.0											0.0			
EMX1 off target 4	G	A	G	T	C	C	T	A	G	C	A	G	G	A	G	A	A	G	A	A	G	A	G	390
untreated					0.0	0.0				0.0														
BE1					0.2	0.2				0.0														
BE2					0.5	0.5				0.0														
BE3					2.2	2.2				0.0														
EMX1 off target 6	G	A	G	T	C	C	G	G	G	A	A	G	G	A	G	A	A	G	A	A	A	G	G	216
untreated					0.0	0.1																		
BE1					0.1	0.1																		
BE2					0.2	0.3																		
BE3					1.0	1.0																		
EMX1 off target 7	G	A	G	C	C	G	G	A	G	C	A	G	A	A	G	A	A	G	G	A	G	G	G	143
untreated				0.0	0.0					0.0														
BE1				0.0	0.0					0.0														
BE2				0.1	0.0					0.0														
BE3				0.1	0.1					0.0														
EMX1 off target 8	A	A	G	T	C	C	G	A	G	G	A	G	A	G	G	A	A	G	A	A	A	G	G	102
untreated					0.0	0.0																		
BE1					0.0	0.0																		
BE2					0.0	0.0																		
BE3					0.1	0.1																		
EMX1 off target 9	G	A	A	T	C	C	A	A	G	C	A	G	G	A	G	A	A	G	A	A	G	G	A	67
untreated					0.0	0.1				0.0														
BE1					0.0	0.1				0.0														
BE2					0.0	0.1				0.0														
BE3					0.3	0.3				0.0														
EMX1 off target 10	A	C	G	T	C	T	G	A	G	C	A	G	A	A	G	A	A	G	A	A	T	G	G	38
untreated		0.0			0.1					0.0														
BE1		0.0			0.3					0.0														
BE2		0.0			0.5					0.0														
BE3		0.0			2.1					0.0														

Supplementary Table 2. Activities of BE1, BE2, and BE3 at FANCF off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the FANCF sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 off-target loci for the FANCF sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 19). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers for each sequence.

																						GUIDE-seq counts
FANCF on target	G	G	A	A	T	C	C	C	T	T	C	T	G	C	A	G	C	A	C	C	T	G	G	4816
untreated						0.0	0.0	0.0			0.0			0.0			0.0		0.0	0.0				
BE1						3.7	3.2	3.4			2.4			0.0			0.0		0.0	0.0				
BE2						4.1	3.5	3.5			0.4			0.0			0.0		0.0	0.0				
BE3						19.1	16.0	16.7			8.8			0.1			0.0		0.0	0.0				
FANCF off target 1	G	G	A	A	C	C	C	C	G	T	C	T	G	C	A	G	C	A	C	C	A	G	G	2099
untreated					0.0	0.0	0.0	0.0			0.0			0.0			0.0		0.0	0.0				
BE1					0.0	0.4	0.0	0.1			0.0			0.0			0.0		0.0	0.0				
BE2					0.2	0.2	0.3	0.3			0.0			0.1			0.0		0.0	0.0				
BE3					1.9	2.1	1.5	1.3			1.3			0.0			0.0		0.0	0.0				
FANCF off target 2	G	G	A	G	T	C	C	C	T	C	C	T	A	C	A	G	C	A	C	C	A	G	G	524
untreated						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE1						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE2						0.1	0.1	0.1		0.1	0.1			0.0			0.0		0.0	0.0				
BE3						0.2	0.2	0.1		0.1	0.1			0.0			0.0		0.0	0.0				
FANCF off target 3	A	G	A	G	G	C	C	C	C	T	C	T	G	C	A	G	C	A	C	C	A	G	G	150
untreated						0.0	0.1	0.1	0.0		0.0			0.0			0.0		0.0	0.0				
BE1						0.0	0.1	0.1	0.0		0.0			0.0			0.0		0.0	0.0				
BE2						0.0	0.1	0.1	0.0		0.0			0.0			0.1		0.0	0.0				
BE3						0.0	0.0	0.0	0.0		0.0			0.0			0.0		0.0	0.0				
FANCF off target 4	A	C	C	A	T	C	C	C	T	C	C	T	G	C	A	G	C	A	C	C	A	G	G	125
untreated		0.0	0.0			0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE1		0.0	0.0			0.0	0.0	0.1		0.1	0.0			0.0			0.0		0.0	0.2				
BE2		0.0	0.0			0.1	0.1	0.1		0.2	0.0			0.0			0.0		0.0	0.0				
BE3		0.0	0.0			0.7	0.4	0.3		0.1	0.0			0.0			0.0		0.0	0.0				
FANCF off target 5	G	G	A	T	T	G	C	C	A	T	C	C	G	C	A	G	C	A	C	C	T	G	G	101
untreated							0.1	0.0			0.0	0.0		0.0			0.0		0.0	0.0				
BE1							0.1	0.0			0.0	0.1		0.0			0.0		0.0	0.0				
BE2							0.1	0.0			0.0	0.0		0.0			0.0		0.0	0.0				
BE3							0.1	0.0			0.0	0.0		0.0			0.0		0.0	0.0				
FANCF off target 6	T	G	A	A	T	C	C	C	A	T	C	T	C	C	A	G	C	A	C	C	A	G	G	78
untreated						0.0	0.0	0.0			0.0		0.1	0.1			0.0		0.0	0.0				
BE1						0.0	0.0	0.0			0.0		0.0	0.0			0.0		0.0	0.0				
BE2						0.2	0.2	0.2			0.2		0.1	0.0			0.0		0.0	0.0				
BE3						0.3	0.3	0.3			0.3		0.1	0.0			0.0		0.0	0.0				
FANCF off target 7	G	G	A	G	T	C	C	C	T	C	C	T	A	C	A	G	C	A	C	C	A	G	G	77
untreated						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE1						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE2						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE3						0.5	0.5	0.5		0.3	0.1			0.0			0.0		0.0	0.0				
FANCF off target 8	G	G	A	G	T	C	C	C	T	C	C	T	G	C	A	G	C	A	C	C	T	G	A	4
untreated						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE1						0.0	0.0	0.0		0.0	0.0			0.0			0.0		0.0	0.0				
BE2						1.4	0.5	0.5		0.4	0.5			0.0			0.0		0.0	0.0				
BE3						0.1	0.1	0.0		0.0	0.0			0.0			0.0		0.0	0.0				

Supplementary Table 3. Activities of BE1, BE2, and BE3 at HEK293 site 2 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 2 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 and dCas9 off-target loci for the HEK293 site 2 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

																							GUIDE-seq counts/ ChIP-seq intensity
HEK site 2 on target	G	A	A	C	A	C	A	A	A	G	C	A	T	A	G	A	C	T	G	C	G	G	G	1670/ 117
untreated				0.0		0.0					0.0						0.0			0.0				
BE1				1.8		2.9					0.0						0.0			0.0				
BE2				8.5		10.5					0.0						0.0			0.0				
BE3				17.6		14.9					0.0						0.0			0.1				
HEK site 2 GUIDE-seq off target 1	G	A	A	C	A	C	A	A	T	G	C	A	T	A	G	A	T	T	G	C	C	G	G	99
untreated				0.0		0.0					0.0									0.1	0.0			
BE1				0.0		0.0					0.1									0.1	0.2			
BE2				0.0		0.4					0.0									0.1	0.0			
BE3				0.2		0.6					0.0									0.1	0.0			
HEK site 2 GUIDE-seq off target 2	A	A	A	C	A	T	A	A	A	G	C	A	T	A	G	A	C	T	G	C	A	A	A	4
untreated				0.0							0.0						0.0			0.0				
BE1				0.0							0.0						0.0			0.0				
BE2				0.0							0.0						0.0			0.0				
BE3				0.0							0.0						0.0			0.0				
HEK site 2 ChIP-seq off target 1	T	C	A	G	G	G	T	G	A	G	C	A	T	A	G	A	C	T	G	C	C	G	G	31
untreated		0.0									0.0						0.0			0.0	0.0			
BE1		0.0									0.0						0.0			0.1	0.0			
BE2		0.0									0.0						0.0			0.1	0.0			
BE3		0.0									0.0						0.0			0.1	0.0			
HEK site 2 ChIP-seq off target 2	G	A	A	T	C	G	G	G	A	G	G	G	G	A	G	A	C	T	G	C	G	G	G	28
untreated					0.0												0.0			0.0				
BE1					0.0												0.0			0.1				
BE2					0.0												0.0			0.1				
BE3					0.0												0.0			0.1				
HEK site 2 ChIP-seq off target 3	T	G	A	A	G	T	G	T	T	G	C	A	T	A	G	A	C	T	G	C	A	G	G	23
untreated											0.0						0.0			0.0				
BE1											0.0						0.0			0.1				
BE2											0.0						0.0			0.0				
BE3											0.0						0.0			0.0				
HEK site 2 ChIP-seq off target 4	G	G	A	G	A	G	A	G	A	G	C	A	T	A	G	A	C	T	G	C	T	G	G	16
untreated											0.0						0.0			0.0				
BE1											0.0						0.0			0.0				
BE2											0.0						0.0			0.0				
BE3											0.0						0.0			0.0				
HEK site 2 ChIP-seq off target 5	C	C	A	A	A	C	A	A	A	A	C	A	T	A	G	A	C	T	G	C	T	G	G	10
untreated	0.0	0.0				0.0					0.0						0.0			0.0				
BE1	0.0	0.0				0.0					0.0						0.0			0.0				
BE2	0.0	0.0				0.0					0.0						0.0			0.0				
BE3	0.0	0.0				0.0					0.0						0.0			0.2				

Supplementary Table 4. Activities of BE1, BE2, and BE3 at HEK293 site 3 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 3 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 off-target loci and the top five known dCas9 off-target loci for the HEK293 site 3 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

																							GUIDE-seq counts/ ChIP-seq intensity
HEK site 3 on target	G	G	C	C	C	A	G	A	C	T	G	A	G	C	A	C	G	T	G	A	T	G	G	2074/ 163
untreated			0.1	0.1	0.1				0.0					0.0		0.0								
BE1			0.1	4.8	4.3				0.0					0.0		0.0								
BE2			3.6	21.3	20.4				0.0					0.0		0.0								
BE3			0.5	33.4	33.1				0.5					0.0		0.2								
HEK site 3 GUIDE-seq off target 1	C	A	C	C	C	A	G	A	C	T	G	A	G	C	A	C	G	T	G	C	T	G	G	327
untreated	0.0		0.0	0.0	0.0				0.0					0.0		0.0				0.0				
BE1	0.0		0.0	0.0	0.0				0.0					0.0		0.0				0.0				
BE2	0.0		0.0	0.0	0.0				0.0					0.0		0.0				0.0				
BE3	0.0		0.0	0.0	0.0				0.0					0.0		0.0				0.0				
HEK site 3 GUIDE-seq off target 2	G	A	C	A	C	A	G	A	C	T	G	G	G	C	A	C	G	T	G	A	G	G	G	306
untreated			0.0		0.0				0.0					0.0		0.0								
BE1			0.0		0.0				0.0					0.0		0.0								
BE2			0.0		0.0				0.0					0.0		0.0								
BE3			0.0		0.0				0.0					0.0		0.0								
HEK site 3 GUIDE-seq off target 3	A	G	C	T	C	A	G	A	C	T	G	A	G	C	A	A	G	T	G	A	G	G	G	136
untreated			0.0		0.0				0.0					0.0										
BE1			0.0		0.0				0.0					0.0										
BE2			0.0		0.0				0.0					0.0										
BE3			0.0		0.9				0.0					0.0										
HEK site 3 GUIDE-seq off target 4	A	G	A	C	C	A	G	A	C	T	G	A	G	C	A	A	G	A	G	A	G	G	G	10
untreated				0.0	0.0				0.0					0.0										
BE1				0.0	0.0				0.0					0.1										
BE2				0.0	0.0				0.0					0.0										
BE3				0.0	0.0				0.0					0.0										
HEK site 3 GUIDE-seq off target 5	G	A	G	C	C	A	G	A	A	T	G	A	G	C	A	C	G	T	G	A	G	G	G	2
untreated				0.0	0.0									0.0		0.0								
BE1				0.0	0.0									0.0		0.0								
BE2				0.0	0.0									0.0		0.0								
BE3				0.0	0.0									0.0		0.0								
HEK site 3 ChIP-seq off target 1	C	A	G	G	A	A	G	C	T	G	G	A	G	C	A	C	G	T	G	A	G	G	G	47
untreated	0.0							0.0						0.0		0.0								
BE1	0.0							0.0						0.0		0.0								
BE2	0.0							0.0						0.0		0.0								
BE3	0.0							0.0						0.0		0.0								
HEK site 3 ChIP-seq off target 2	A	A	G	G	C	T	G	A	G	G	G	A	G	C	A	C	G	T	G	A	A	G	G	46
untreated					0.0									0.0		0.0								
BE1					0.0									0.0		0.0								
BE2					0.0									0.0		0.0								
BE3					0.0									0.0		0.0								
HEK site 3 ChIP-seq off target 3	G	T	C	A	G	G	G	G	A	A	G	A	G	C	A	C	G	T	G	A	C	G	G	33
untreated			0.0											0.0		0.0					0.0			
BE1			0.0											0.0		0.0					0.0			
BE2			0.0											0.0		0.0					0.0			
BE3			0.0											0.0		0.0					0.0			
HEK site 3 ChIP-seq off target 4	G	T	T	G	T	G	A	A	C	T	G	A	G	C	A	C	G	T	G	A	G	G	G	31
untreated									0.0					0.0		1.7								
BE1									0.0					0.0		0.7								
BE2									0.0					0.0		0.4								
BE3									0.0					0.0		0.4								
HEK site 3 ChIP-seq off target 5	A	T	A	T	T	T	G	C	T	G	G	A	G	C	A	C	G	T	G	A	A	G	G	31
untreated								0.0						0.0		0.0								
BE1								0.0						0.0		0.0								
BE2								0.0						0.0		0.0								
BE3								0.0						0.0		0.0								

Supplementary Table 5. Activities of BE1, BE2, and BE3 at HEK293 site 4 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 4 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus the top ten known Cas9 off-target loci and the top five known dCas9 off-target loci for the HEK293 site 4 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

																							GUIDE-seq counts/ ChIP-seq intensity
HEK site 4 on target	G	G	C	A	C	T	G	C	G	G	C	T	G	G	A	G	G	T	G	G	G	G	1054/ 49
untreated			0.0		0.0			0.1			0.0												
BE1			0.0		0.4			0.1			0.0												
BE2			0.1		12.7			0.1			0.0												
BE3			1.5		22.3			1.0			0.0												
HEK site 4 GUIDE-seq off target 1	T	G	C	A	C	T	G	C	G	G	C	C	G	G	A	G	G	A	G	G	T	G	G	2475
untreated			0.0		0.0			0.0			0.0	0.0											
BE1			0.0		0.6			0.1			0.1	0.0											
BE2			0.3		3.3			0.3			0.0	0.0											
BE3			0.2		10.2			0.3			0.0	0.0											
HEK site 4 GUIDE-seq off target 2	G	G	C	T	C	T	G	C	G	G	C	T	G	G	A	G	G	G	G	G	T	G	G	2020
untreated			0.0		0.0			0.1			0.0												
BE1			0.0		0.6			0.1			0.0												
BE2			0.0		0.4			0.1			0.1												
BE3			0.0		3.3			0.1			0.0												
HEK site 4 GUIDE-seq off target 3	G	G	C	A	C	G	A	C	G	G	C	T	G	G	A	G	G	T	G	G	G	G	G	1375
untreated			0.0		0.0			0.0			0.0												
BE1			0.1		11.8			4.0			0.0												
BE2			0.0		9.6			3.9			0.0												
BE3			0.1		19.0			1.9			0.1												
HEK site 4 GUIDE-seq off target 4	G	G	C	A	T	C	A	C	G	G	C	T	G	G	A	G	G	T	G	G	A	G	G	1097
untreated			0.0		0.0			0.0			0.0												
BE1			0.0		0.2			0.3			0.0												
BE2			0.5		5.3			2.7			0.0												
BE3			0.2		18.1			3.1			0.0												
HEK site 4 GUIDE-seq off target 5	G	G	C	G	C	T	G	C	G	G	C	G	G	G	A	G	G	T	G	G	A	G	G	1002
untreated			0.0		0.0			0.1			0.0												
BE1			0.0		0.0			0.0			0.0												
BE2			0.0		0.0			0.1			0.0												
BE3			0.1		0.1			0.1			0.0												
HEK site 4 GUIDE-seq off target 6	G	G	C	A	C	T	G	A	G	A	C	T	G	G	G	G	G	T	G	G	G	G	G	982
untreated			0.0		0.0						0.0												
BE1			0.0		0.5						0.3												
BE2			0.8		3.6						0.0												
BE3			0.0		6.8						0.0												
HEK site 4 GUIDE-seq off target 7	A	G	C	A	G	T	G	C	G	G	C	T	A	G	A	G	G	T	G	G	T	G	G	981
untreated			0.0					0.1			0.0												
BE1			0.0					0.1			0.0												
BE2			0.0					0.1			0.0												
BE3			0.0					0.2			0.0												
HEK site 4 GUIDE-seq off target 8	G	G	C	A	C	T	G	C	T	A	C	T	G	G	G	G	G	T	G	G	T	G	G	789
untreated			0.0		0.0			0.0			0.0												
BE1			0.0		2.1			0.0			0.0												
BE2			0.2		5.1			0.5			0.0												
BE3			0.2		8.2			0.1			0.0												
HEK site 4 GUIDE-seq off target 9	G	G	C	A	C	T	G	G	G	G	C	T	G	G	G	G	G	A	G	G	G	G	G	712
untreated			0.0		0.0						0.0												
BE1			0.0		0.3						0.0												
BE2			0.0		0.4						0.0												
BE3			0.0		0.9						0.0												
HEK site 4 GUIDE-seq off target 10	G	G	C	A	C	T	G	G	G	G	T	T	G	G	A	G	G	T	G	G	G	G	G	664
untreated			0.0		0.0																		
BE1			0.0		0.0																		
BE2			0.6		3.5																		
BE3			0.3		10.0																		


HEK site 4 ChIP-seq off target 1
G T G G C T G G A G G T G G A G G T G G G G G
110
untreated 0.0
BE1 0.0
BE2 0.0
BE3 0.0
HEK site 4 ChIP-seq off target 3
G A G G G A A G G G C T G G A G G T G G A G G
89
untreated 0.0
BE1 0.0
BE2 0.0
BE3 0.0

Supplementary Table 6: Mutation rates of non-protospacer bases following BE3-mediated correction of the Alzheimer's disease-associated *APOE4* allele to *APOE3r* in mouse astrocytes. The DNA sequence of the 50 bases on either side of the protospacer from Fig. 4a and Extended Data Fig. 7a is shown with each base's position relative to the protospacer. The side of the protospacer distal to the PAM is designated with positive numbers, while the side that includes the PAM is designated with negative numbers, with the PAM shown in blue. Underneath each sequence are the percentages of total DNA sequencing reads with the corresponding base for untreated cells, for cells treated with BE3 and an sgRNA targeting the *APOE4* C158R mutation, or for cells treated with BE3 and an sgRNA targeting the *VEGFA* locus. Neither BE3-treated sample resulted in mutation rates above those of untreated controls.

APOE4	C50	A49	C48	C47	T46	G45	C44	G43	C42	A41	A40	G39	C38	T37	G36	C35	G34	T33	A32	A31	G30	C29	G28	G27	C26
Untreated																									
A	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	100±0.1
G	0.1±0.1	0.1±0.1	0±0	0±0	0.1±0.1	100±0.1	0±0	99.9±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0±0	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0±0
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + on-target sgRNA																									
A	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0±0	0.1±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0±0	0±0	100±0.1
G	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + off-target sgRNA																									
A	0±0	100±0.1	0±0	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0±0	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0
C	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	100±0	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1
G	0±0	0.1±0.1	0±0	0±0	0±0	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	100±0.1	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	0±0	100±0.1	100±0.1	0±0
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0±0	0±0	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1

APOE4	T25	C24	C23	T22	C21	C20	G19	C18	G17	A16	T15	G14	C13	C12	G11	A10	T9	G8	A7	C6	C5	T4	G3	C2	A1
Untreated																									
A	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1
C	0.1±0.1	100±0	100±0.1	0.1±0.1	100±0.1	100±0.1	0±0	100±0.1	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	100±0.1	0±0	0±0	0.1±0.1	0±0	0±0	100±0.1	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1
G	0±0	0±0	0±0	0.1±0.1	0±0	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0±0	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0±0	0±0	100±0.1	0±0	0.1±0.1
T	100±0.1	0±0	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1
BE3 + on-target sgRNA																									
A	0±0	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0±0	0.1±0.1	0.2±0.1	100±0.1	0.1±0.1	0.2±0.1	99.9±0.2	0±0	0.1±0.1	0.2±0.2	0.1±0.1	0.1±0.1	99.9±0.2
C	0.1±0.1	99.9±0.1	100±0.1	0.1±0.1	99.5±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0±0	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.2±0.2
G	0±0	0±0	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	99.9±0.1	0.1±0.1	0±0	99.9±0.1	0.2±0.2	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1
T	100±0.1	0.2±0.1	0.1±0.1	100±0.1	0.5±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	99.9±0.2	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + off-target sgRNA																									
A	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0±0	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
C	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0±0	100±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0±0	0±0	0±0	0.1±0.1	0±0	0.1±0.1	100±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0±0	0±0	0±0	100±0.1	0.1±0.1	0.1±0.1
T	100±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0±0

APOE4	A-1	G-2	G-3	C-4	C-5	G-6	G-7	G-8	G-9	C-10	C-11	C-12	G-13	C-14	G-15	A-16	G-17	G-18	G-19	C-20	G-21	C-22	C-23	G-24	A-25
Untreated																									
A	100±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.2±0.1	0.1±0.1	0±0	0.1±0.1	100±0.1
C	0.1±0.1	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	99.9±0.1	0.1±0.1	0.1±0.1	0±0	0±0	0±0	100±0.1	0.1±0.1	100±0.1	100±0.1	0±0	0.1±0.1
G	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0±0	99.9±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1	99.9±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	99.9±0.1	0.1±0.1	0.1±0.1	100±0.2	0.1±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.2	0.1±0.1
BE3 + on-target sgRNA																									
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.2±0.1	0.2±0.2	0.2±0.1	0.2±0.1	0.1±0.1	0.1±0.1	99.9±0.1
C	0.1±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0±0	0±0	0±0	0.1±0.1	0±0	99.8±0.2	0±0	99.9±0.1	100±0.1	0±0	0±0
G	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	99.9±0.1	0±0	100±0.1	0.1±0.1	100±0.1	100±0.1	99.9±0.1	0.2±0.1	99.8±0.2	0±0	0±0	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.2	0.1±0.1	0.1±0.1	0.2±0.1
BE3 + off-target sgRNA																									
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.2±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
C	0.1±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0±0	0±0	0±0	100±0.1	100±0.1	100±0.1	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0±0	0±0	100±0.1	0.1±0.1	100±0.1	100±0.1	0±0	0±0
G	0.1±0.1	100±0.1	100±0.1	0±0	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0±0	0±0	0±0	99.9±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	99.9±0.1	0±0	0±0	100±0.1	0±0
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0±0	0.1±0.1	0±0	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1

APOE4	G-26	C-27	G-28	C-29	G-30	G-31	C-32	C-33	T-34	C-35	A-36	G-37	C-38	G-39	C-40	C-41	A-42	T-43	C-44	C-45	G-46	C-47	G-48	A-49	G-50
Untreated																									
A	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.2±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	0±0	100±0.1	0.1±0.1	99.9±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	99.8±0.1	0.1±0.1	100±0.1	100±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1
G	100±0.1	0.1±0.1	99.9±0.2	0.1±0.1	100±0.1	100±0.1	0±0	0±0	0±0	0±0	0.1±0.1	100±0.1	0.1±0.1	99.9±0.1	0±0	0±0	0.1±0.1	0.5±0.1	0.1±0.1	0.1±0.1	99.9±0.2	0±0	99.7±0.1	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.2±0.2	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.3±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	99.6±0.1	0.1±0.1	0.1±0.1	0.2±0.2	0.1±0.1	0.3±0.1	0±0	0.1±0.1
BE3 + on-target sgRNA																									
A	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.2±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	0±0	100±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	100±0.1	99.9±0.1	0±0	0±0	0±0	99.7±0.1	0.1±0.1	100±0.1	100±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	0.2±0.2
G	100±0.1	0.1±0.1	100±0.1	0±0	100±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1	0±0	0.1±0.1	100±0.1	0.1±0.1	99.8±0.2	0.1±0.1	0±0	0.1±0.1	0.5±0.5	0±0	0±0	100±0.1	0.1±0.1	99.6±0.4	0.1±0.1	99.9±0.2
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	100±0.1	0.2±0.1	0.1±0.1	0.1±0.1	0.4±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	99.6±0.5	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.4±0.3	0±0	0±0
BE3 + off-target sgRNA																									
A	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0±0	0±0	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	0.1±0.1	100±0.1	0±0	100±0.1	0.1±0.1	0±0	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	99.8±0.1	0.1±0.1	100±0.1	100±0.1	0±0	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0±0	0.1±0.1
G	100±0.1	0.1±0.1	100±0.1	0±0	100±0.1	100±0.1	0±0	0±0	0±0	0±0	0.1±0.1	100±0.1	0.1±0.1	99.9±0.1	0±0	0±0	0.1±0.1	0.4±0.1	0±0	0±0	100±0.1	0±0	99.7±0.2	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0±0	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.2	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	99.7±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.4±0.2	0±0	0.1±0.1

Supplementary Table 7: Mutation rates of non-protospacer bases following BE3-mediated correction of the cancer-associated p53 Y163C mutation in HCC1954 human cells. The DNA sequence of the 50 bases on either side of the protospacer from Fig. 4b and Extended Data Fig. 7b is shown with each base's position relative to the protospacer. The side of the protospacer distal to the PAM is designated with positive numbers, while the side that includes the PAM is designated with negative numbers, with the PAM shown in blue. Underneath each sequence are the percentages of total sequencing reads with the corresponding base for untreated cells, for cells treated with BE3 and an sgRNA targeting the *TP53* Y163C mutation, or for cells treated with BE3 and an sgRNA targeting the *VEGFA* locus. Neither BE3-treated sample resulted in mutational rates above those of untreated controls.

TP53	C-50	C-49	T-48	G-47	T-46	G-45	C-44	A-43	G-42	C-41	T-40	G-39	T-38	G-37	G-36	G-35	T-34	T-33	G-32	A-31	T-30	T-29	C-28	C-27	A-26
Untreated																									
A	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
C	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + on-target sgRNA																									
A	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
C	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	99.8±0.3	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.3±0.3	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1
BE3 + off-target sgRNA																									
A	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
C	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1

TP53	C-25	A-24	C-23	C-22	C-21	C-20	C-19	G-18	C-17	C-16	C-15	G-14	G-13	C-12	A-11	C-10	C-9	C-8	G-7	C-6	G-5	T-4	C-3	C-2	G-1
Untreated																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + on-target sgRNA																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	99.9±0.2	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.2	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + off-target sgRNA																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.5±0.7	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	99.6±0.6	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1
G	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1

TP53	A1	G2	T3	C4	A5	C6	A7	G8	C9	A10	C11	A12	T13	G14	A15	C16	G17	G18	A19	G20	G21	T22	T23	G24	T25
Untreated																									
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
G	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1
BE3 + on-target sgRNA																									
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.3±0.4	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.5±0.7	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
G	0.1±0.1	99.9±0.3	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	99.8±0.4	0.1±0.1	100±0.1	99.6±0.7	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1
BE3 + off-target sgRNA																									
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
G	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1

TP53	G26	A27	G28	G29	C30	G31	C32	T33	G34	C35	C36	C37	C38	C39	A40	C41	C42	A43	T44	G45	A46	G47	C48	G49	C50
Untreated																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1
G	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + on-target sgRNA																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.2±0.3	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	99.9±0.3	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1
G	100±0.1	0.1±0.1	99.7±0.6	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	0.4±0.5	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
BE3 + off-target sgRNA																									
A	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	100±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1
G	100±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1

Supplementary Table 8: List of 911 base-editable gene variants associated with human disease with an NGG PAM positioned appropriately. Shown for each gene variant are, from left to right, the Single Nucleotide Polymorphism Database (dbSNP) identification number, the genotype (written as the NCBI GenBank identification number of the gene, the gene name, the chromosome location and DNA base substitution of the SNP, and the amino acid substitution caused by the SNP), the protospacer and PAM sequence(s) that would be used in combination with a base editor to correct each SNP (shown as the coding strand sequence), and the associated genetic disease. The activity window was defined as positions 4-8. Variants containing only a single C within the activity window are highlighted in yellow (284 variants). We note that Cas9 variants with altered PAM specificities may enable many disease-associated gene variants not listed below to be addressed by base editing.

dbSNP #	Genotype	Protospacer and PAM sequence(s)	Associated genetic disease
755445790	NM_000391.3(TPP1):c.887-10A>G	TTTYTTTTTTTTTTTTTTTGAGG	Ceroid lipofuscinosis, neuronal, 2
113994167	NM_000018.3(ACADVL):c.848T>C (p.Val283Ala)	TTTGYGGTGGAGAGGGGCTTCGG, TTGYGGTGGAGAGGGGCTTCGGG	Very long chain acyl-CoA dehydrogenase deficiency
119470018	NM_024996.5(GFM1):c.521A>G (p.Asn174Ser)	TTGYTAATAAAAGTTAGAAACGG	Combined oxidative phosphorylation deficiency 1
115650537	NM_000426.3(LAMA2):c.8282T>C (p.Ile2761Thr)	TTGAYAGGGAGCAAGCAGTTCGG, TGAYAGGGAGCAAGCAGTTCGGG	Merosin deficient congenital muscular dystrophy
587777752	NM_014946.3(SPAST):c.1688-2A>G	TTCYGTAAAACATAAAAGTCAGG	Spastic paraplegia 4, autosomal dominant
794726821	NM_001165963.1(SCN1A):c.4055T>C (p.Leu1352Pro)	TTCYGGTTTGTCTTATATTCTGG	Severe myoclonic epilepsy in infancy
397514745	NM_001130089.1(KARS):c.517T>C (p.Tyr173His)	CTTCYATGATCTTCGAGGAGAGG, TTCYATGATCTTCGAGGAGAGGG	Deafness, autosomal recessive 89
376960358	NM_001202.3(BMP4):c.362A>G (p.His121Arg)	TTCGTGGYGGAAGCTCCTCACGG	Microphthalmia syndromic 6
606231280	NM_001287223.1(SCN11A):c.1142T>C (p.Ile381Thr)	CTTCAYTGTGGTCATTTTCCTGG, TTCAYTGTGGTCATTTTCCTGGG	Episodic pain syndrome, familial, 3
387906735	m.608A>G	TTCAGYGTATTGCTTTGAGGAGG	
199474663	m.3260A>G	TTAAGTTYTATGCGATTACCGGG	Cardiomyopathy with or without skeletal myopathy
104894962	NM_003413.3(ZIC3):c.1213A>G (p.Lys405Glu)	TGTGTTYGCGCAGGGAGCTCGGG, ATGTGTTYGCGCAGGGAGCTCGG	Heterotaxy, visceral, X-linked
796053181	NM_021007.2(SCN2A):c.1271T>C (p.Val424Ala)	TGTGGYGGCCATGGCCTATGAGG	not provided
267606788	NM_000129.3(F13A1):c.728T>C (p.Met243Thr)	TGTGAYGGACAGAGCACAAATGG	Factor xiii, a subunit, deficiency of
397514503	NM_003863.3(DPM2):c.68A>G (p.Tyr23Cys)	TGTAGYAGGTGAAGATGATCAGG	Congenital disorder of glycosylation type 1u
104893973	NM_000416.2(IFNGR1):c.260T>C (p.Ile87Thr)	TGTAATAYTTCTGATCATGTTGG	Disseminated atypical mycobacterial infection, Mycobacterium tuberculosis, susceptibility to
121908466	NM_005682.6(ADGRG1):c.263A>G (p.Tyr88Cys)	TGGYAGAGGCCCCTGGGGTCAGG	Polymicrogyria, bilateral frontoparietal
147952488	NM_002437.4(MPV17):c.186+2T>C	TGGYAAGTTCTCCCCTCAACAGG	Navajo neurohepatopathy
121909537	NM_001145.4(ANG):c.121A>G (p.Lys41Glu)	TGGTTYGGCATCATAGTGCTGGG, GTGGTTYGGCATCATAGTGCTGG	Amyotrophic lateral sclerosis type 9
121918489	NM_000141.4(FGFR2):c.1018T>C (p.Tyr340His)	TGGGGAAYATACGTGCTTGGCGG, GGGGAAYATACGTGCTTGGCGGG	Crouzon syndrome
121434463	m.12320A>G	GAGTYGCACCAAAATTTTTGGGG, GGAGTYGCACCAAAATTTTTGGG, TGGAGTYGCACCAAAATTTTTGG	Mitochondrial myopathy
121908046	NM_000403.3(GALE):c.101A>G (p.Asn34Ser)	TGGAAGYTATCGATGACCACAGG	UDPglucose-4-epimerase deficiency
431905512	NM_003764.3(STX11):c.173T>C (p.Leu58Pro)	TGCYGGTGGCCGACGTGAAGCGG	Hemophagocytic lymphohistiocytosis, familial, 4
121917905	NM_000124.3(ERCC6):c.2960T>C (p.Leu987Pro)	TGCYAAAAGACCCAAAACAAAGG	Cerebro-oculo-facio-skeletal syndrome
121918500	NM_000141.4(FGFR2):c.874A>G (p.Lys292Glu)	TGCTYGATCCACTGGATGTGGGG, GTGCTYGATCCACTGGATGTGGG, CGTGCTYGATCCACTGGATGTGG	Crouzon syndrome
60431989	NM_000053.3(ATP7B):c.3443T>C (p.Ile1148Thr)	TGCTGAYTGGAAACCGTGAGTGG	Wilson disease
78950939	NM_000250.1(MPO):c.518A>G (p.Tyr173Cys)	GTGCGGYATTTGTCCTGCTCCGG, TGCGGYATTTGTCCTGCTCCGGG	Myeloperoxidase deficiency
115677373	NM_201631.3(TGM5):c.763T>C (p.Trp255Arg)	TGCGGAGYGGACGGGCAGCGTGG	Peeling skin syndrome, acral type
5030804	NM_000551.3(VHL):c.233A>G (p.Asn78Ser)	GCGAYTGCAGAAGATGACCTGGG, TGCGAYTGCAGAAGATGACCTGG	Von Hippel-Lindau syndrome
397508328	NM_000492.3(CFTR):c.1A>G (p.Met1Val)	GCAYGGTCTCTCGGGCGCTGGGG, TGCAYGGTCTCTCGGGCGCTGGG,	Cystic fibrosis

		CTGCAYGGTCTCTCGGGCGCTGG	
137853299	NM_000362.4(TIMP3):c.572A>G (p.Tyr191Cys)	TGCAGYAGCCGCCCTTCTGCCGG	Sorsby fundus dystrophy
121908549	NM_000334.4(SCN4A):c.3478A>G (p.Ile1160Val)	TGAYGGAGGGGATGGCGCCTAGG	
121909337	NM_001451.2(FOXF1):c.1138T>C (p.Ter380Arg)	TGATGYGAGGCTGCCGCCGCAGG	Alveolar capillary dysplasia with misalignment of pulmonary veins
281875320	NM_005359.5(SMAD4):c.1500A>G (p.Ile500Met)	TGAGYATGCATAAGCGACGAAGG	Myhre syndrome
730880132	NM_170707.3(LMNA):c.710T>C (p.Phe237Ser)	TGAGTYTGAGAGCCGGCTGGCGG	Primary dilated cardiomyopathy
281875322	NM_005359.5(SMAD4):c.1498A>G (p.Ile500Val)	TGAGTAYGCATAAGCGACGAAGG	Hereditary cancer-predisposing syndrome, Myhre syndrome
72556283	NM_000531.5(OTC):c.527A>G (p.Tyr176Cys)	TGAGGYAATCAGCCAGGATCTGG	not provided
74315311	NM_020435.3(GJC2):c.857T>C (p.Met286Thr)	TGAGAYGGCCCACCTGGGCTTGG, GAGAYGGCCCACCTGGGCTTGGG	Leukodystrophy, hypomyelinating, 2
121912495	NM_170707.3(LMNA):c.1139T>C (p.Leu380Ser)	TCTYGGAGGGCGAGGAGGAGAGG	Congenital muscular dystrophy, LMNA-related
128620184	NM_000061.2(BTK):c.1288A>G (p.Lys430Glu)	TCTYGATGGCCACGTCGTACTGG	X-linked agammaglobulinemia
118192252	NM_004519.3(KCNQ3):c.1403A>G (p.Asn468Ser)	TCTTTAYTGTTTAAGCCAACAGG	Benign familial neonatal seizures 2, not specified
121909142	NM_001300.5(KLF6):c.190T>C (p.Trp64Arg)	TCTGYGGACCAAAATCATTCTGG	
104895503	NM_001127255.1(NLRP7):c.2738A>G (p.Asn913Ser)	TCTGGYTGATACTCAAGTCCAGG	Hydatidiform mole
587783035	NM_000038.5(APC):c.1744-2A>G	TCCYAGTAAGAAACAGAATATGG	Familial adenomatous polyposis 1
72556289	NM_000531.5(OTC):c.541-2A>G	TCCYAAAAGGCACGGGATGAAGG	not provided
28937313	NM_005502.3(ABCA1):c.2804A>G (p.Asn935Ser)	TCCAYTGTGGCCCAGGAAGGAGG, CGCTCCAYTGTGGCCCAGGAAGG	Tangier disease
143246552	NM_001003811.1(TEX11):c.511A>G (p.Met171Val)	TCCAYGGTCAAGTCAGCCTCAGG, CCAYGGTCAAGTCAGCCTCAGGG	Spermatogenic failure, X-linked, 2
587776451	NM_002049.3(GATA1):c.2T>C (p.Met1Thr)	CTCCAYGGAGTTCCCTGGCCTGG, TCCAYGGAGTTCCCTGGCCTGGG, CCAYGGAGTTCCCTGGCCTGGGG	GATA-1-related thrombocytopenia with dyserythropoiesis
121908403	NM_021102.3(SPINT2):c.488A>G (p.Tyr163Cys)	TCCAYAGATGAAGTTATTGCAGG	Diarrhea 3, secretory sodium, congenital, syndromic
281874738	NM_000495.4(COL4A5):c.438+2T>C	CTCCAGYAAGTTATAAAATTTGG, TCCAGYAAGTTATAAAATTTGGG	Alport syndrome, X-linked recessive
730880279	NM_030653.3(DDX11):c.2271+2T>C	TCCAGGYGCGGGCGTCATGCTGG, CCAGGYGCGGGCGTCATGCTGGG	Warsaw breakage syndrome
28940272	NM_017890.4(VPS13B):c.8978A>G (p.Asn2993Ser)	TCAYTGATAAGCAGGGCCCAGGG, TTCAYTGATAAGCAGGGCCCAGG	Cohen syndrome, not specified
137852375	NM_000132.3(F8):c.5372T>C (p.Met1791Thr)	TCAYGGTGAGTTAAGGACAGTGG	Hereditary factor VIII deficiency disease
11567847	NM_021961.5(TEAD1):c.1261T>C (p.Tyr?His)	TCATATTYACAGGCTTGTAAAGG	
786203989	NM_016069.9(PAM16):c.226A>G (p.Asn76Asp)	CATAGTYCTGCAGAGGAGAGGGG, TCATAGTYCTGCAGAGGAGAGGG	Chondrodysplasia, megarbane-dagher-melki type
587776437	NC_012920.1:m.9478T>C	TCAGAAGYTTTTTTCTTCGCAGG	Leigh disease
121912474	NM_000424.3(KRT5):c.20T>C (p.Val7Ala)	TCAAGTGYGTCCTTCCGGAGCGG, CAAGTGYGTCCTTCCGGAGCGGG, AAGTGYGTCCTTCCGGAGCGGGG, AGTGYGTCCTTCCGGAGCGGGGG	Epidermolysis bullosa simplex, Koebner type
104886461	NM_020533.2(MCOLN1):c.406-2A>G	TACYGTGGGCAGAGAAGGGGAGG, AGGTACYGTGGGCAGAGAAGGGG, CAGGTACYGTGGGCAGAGAAGGG	Ganglioside sialidase deficiency
104894275	NM_000317.2(PTS):c.155A>G (p.Asn52Ser)	TAAYTGTGCCCATGGCCATTTGG	6-pyruvoyl-tetrahydropterin synthase deficiency
587777562	NM_015599.2(PGM3):c.737A>G (p.Asn246Ser)	TAAATGAYTGAGTTTGCCCTTGG	Immunodeficiency 23
121964906	NM_000027.3(AGA):c.916T>C (p.Cys306Arg)	GTTATAYGTGCCAATGTGACTGG	Aspartylglycosaminuria
28941769	NM_000356.3(TCOF1):c.149A>G (p.Tyr50Cys)	GTGTGTAYAGATGTCCAGAAGGG	Treacher collins syndrome 1
121434464	m.12297T>C	GTCYTAGGCCCCAAAAATTTTGG	Cardiomyopathy, mitochondrial
121908407	NM_054027.4(ANKH):c.143T>C (p.Met48Thr)	GTCGAGAYGCTGGCCAGCTACGG, TCGAGAYGCTGGCCAGCTACGGG	Chondrocalcinosis 2
59151893	NM_000422.2(KRT17):c.275A>G (p.Asn92Ser)	GTCAYTGAGGTTCTGCATGGTGG, GCGGTCAYTGAGGTTCTGCATGG	Pachyonychia congenita type 2
121909499	NM_002427.3(MMP13):c.272T>C (p.Met91Thr)	GTCAYGAAAAAGCCAAGATGCGG, TCAYGAAAAAGCCAAGATGCGGG	
61748478	NM_000552.3(VWF):c.2384A>G (p.Tyr795Cys)	GTCAYAGTTCTGGCACGTTTTGG	von Willebrand disease type 2N

387906889	NM_006796.2(AFG3L2):c.1847A>G (p.Tyr616Cys)	GTAYAGAGGTATTGTTCTTTTGG	Spastic ataxia 5, autosomal recessive
118203907	NM_000130.4(F5):c.5189A>G (p.Tyr1730Cys)	GTAGYAGGCCCAAGCCCGACAGG	Factor V deficiency
118203945	NM_013319.2(UBIAD1):c.305A>G (p.Asn102Ser)	GTAAGTGYTGACCAAATTACCGG	Schnyder crystalline corneal dystrophy
267607080	NM_005633.3(SOS1):c.1294T>C (p.Trp432Arg)	GGTYGGGAGGGAAAAGACATTGG	Noonan syndrome 4, Rasopathy
137852953	NM_012464.4(TLL1):c.1885A>G (p.Ile629Val)	GGTTAYGGTGCCGTTAAGTTTGG	Atrial septal defect 6
118203949	NM_013319.2(UBIAD1):c.695A>G (p.Asn232Ser)	GGTGTTGYTGGAATGGAGAATGG	Schnyder crystalline corneal dystrophy
137852952	NM_012464.4(TLL1):c.713T>C (p.Val238Ala)	GGGATTGYTGTTCATGAATTGGG	Atrial septal defect 6
41460449	m.3394T>C	GGCYATATACAACTACGCAAAGG	Leber optic atrophy
80357281	NM_007294.3(BRCA1):c.5291T>C (p.Leu1764Pro)	GGGCYAGAAATCTGTTGCTATGG, GGCYAGAAATCTGTTGCTATGGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
5030764	NM_000174.4(GP9):c.182A>G (p.Asn61Ser)	GGCTGYTGTTGGCCAGCAGAAGG	Bernard-Soulier syndrome type C
72556282	NM_000531.5(OTC):c.526T>C (p.Tyr176His)	GGCTGATYACCTCACGCTCCAGG, GATYACCTCACGCTCCAGGTTGG	not provided
121913594	NM_000530.6(MPZ):c.242A>G (p.His81Arg)	GGCATAGYGGAAGATCTATGAGG	Charcot-Marie-Tooth disease type 1B
587777736	NM_017617.3(NOTCH1):c.1285T>C (p.Cys429Arg)	GGCAAGYGCATCAACACGCTGGG, GGGCAAGYGCATCAACACGCTGG	Adams-Oliver syndrome 1, Adams-Oliver syndrome 5
63750912	NM_016835.4(MAPT):c.1839T>C (p.Asn613=)	GGATAAYATCAAACACGTCCCGG, GATAAYATCAAACACGTCCCGGG	Frontotemporal dementia
121918075	NM_000371.3(TTR):c.401A>G (p.Tyr134Cys)	GGAGYAGGGGCTCAGCAGGGCGG, ATAGGAGYAGGGGCTCAGCAGGG	Amyloidogenic transthyretin amyloidosis
730882063	NM_004523.3(KIF11):c.2547+2T>C	GGAGGYAATAACTTTGTAAGTGG	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
397516156	NM_000257.3(MYH7):c.2546T>C (p.Met849Thr)	GGAGAYGGCCTCCATGAAGGAGG	Primary familial hypertrophic cardiomyopathy, Cardiomyopathy
118204430	NM_000035.3(ALDOB):c.442T>C (p.Trp148Arg)	GGAAGYGGCGTGCTGTGCTGAGG	Hereditary fructosuria
200198778	NM_013382.5(POMT2):c.1997A>G (p.Tyr666Cys)	GGAAGYAGTGGTGGAAGTAGAGG	Congenital muscular dystrophy, Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2, Muscular dystrophy, Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B2
754896795	NM_004006.2(DMD):c.6982A>T (p.Lys2328Ter)	GCTTTTYTTCAAGCTGCCCAAGG	Duchenne muscular dystrophy, Becker muscular dystrophy, Dilated cardiomyopathy 3B
148924904	NM_000546.5(TP53):c.488A>G (p.Tyr163Cys)	GCTTGYAGATGGCCATGGCGCGG	Hereditary cancer-predisposing syndrome
786204770	NM_016035.4(COQ4):c.155T>C (p.Leu52Ser)	GCTGTYGGCCGCCGGCTCCGCGG	COENZYME Q10 DEFICIENCY, PRIMARY, 7
121909520	NM_001100.3(ACTA1):c.350A>G (p.Asn117Ser)	CGGYTGGCCTTGGGATTGAGGGG, GCGGYTGGCCTTGGGATTGAGGG, CGCGGYTGGCCTTGGGATTGAGG	Nemaline myopathy 3
587776879	NM_004656.3(BAP1):c.438-2A>G	GCCYGGGGAAAAACAGAGTCAGG	Tumor predisposition syndrome
727504434	NM_000501.3(ELN):c.890-2A>G	GCCYGAAAACACAGCCACAGAGG	Supravalvar aortic stenosis
119455953	NM_000391.3(TPP1):c.1093T>C (p.Cys365Arg)	GCCGGGYGTTGGTCTGTCTCTGG	Ceroid lipofuscinosis, neuronal, 2
121964983	NM_000481.3(AMT):c.125A>G (p.His42Arg)	GCCAGGYGGAAGTCATAGAGCGG	Non-ketotic hyperglycinemia
121908300	NM_001005741.2(GBA):c.751T>C (p.Tyr251His)	GCCAGAYACTTTGTGAAGTAAGG, CCAGAYACTTTGTGAAGTAAGGG	Gaucher disease, type 1
786205083	NM_003494.3(DYSF):c.3443-33A>G	GCCAGAGYGAGTGGCTGGAGTGG	Limb-girdle muscular dystrophy, type 2B
121908133	NM_175073.2(APTX):c.602A>G (p.His201Arg)	GCCAAYGGTAACGGGCCTTTGGG, AGCCAAYGGTAACGGGCCTTTGG	Adult onset ataxia with oculomotor apraxia
587777195	NM_005017.3(PCYT1A):c.571T>C (p.Phe191Leu)	GCATGYTTGCTCCAACACAGAGG	Spondylometaphyseal dysplasia with cone-rod dystrophy
431905520	NM_014714.3(IFT140):c.4078T>C (p.Cys1360Arg)	CAAGCAGYGTGAGCTGCTCCTGG, GCAGYGTGAGCTGCTCCTGGAGG	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia
121912889	NM_001844.4(COL2A1):c.4172A>G (p.Tyr1391Cys)	GCAGTGGYAGGTGATGTTCTGGG	Spondyloperipheral dysplasia, Platyspondylic lethal skeletal dysplasia Torrance type
137854492	NM_001363.4(DKC1):c.1069A>G (p.Thr357Ala)	GCAGGYAGAGATGACCGCTGTGG	Dyskeratosis congenita X-linked
121434362	NM_152783.4(D2HGDH):c.1315A>G (p.Asn439Asp)	GCAGGTYACCATCTCCTGGAGGG, TGCAGGTYACCATCTCCTGGAGG	D-2-hydroxyglutaric aciduria 1
80338732	NM_002764.3(PRPS1):c.344T>C (p.Met115Thr)	GCAAATAYGCTATCTGTAGCAGG	Charcot-Marie-Tooth disease, X-linked recessive, type 5
387906675	NM_000313.3(PROS1):c.701A>G (p.Tyr234Cys)	GATTAYATCTGTAGCCTTCGGGG, AGATTAYATCTGTAGCCTTCGGG, GAGATTAYATCTGTAGCCTTCGG	Thrombophilia due to protein S deficiency, autosomal recessive

28935478	NM_000061.2(BTK):c.1082A>G (p.Tyr361Cys)	GATGGYAGTTAATGAGCTCAGGG, TGATGGYAGTTAATGAGCTCAGG	
201777056	NM_005050.3(ABCD4):c.956A>G (p.Tyr319Cys)	GATGAGGYAGATGCACACAAAGG	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE
121918528	NM_000098.2(CPT2):c.359A>G (p.Tyr120Cys)	GATAGGYACATATCAAACCAGGG, AGATAGGYACATATCAAACCAGG	Carnitine palmitoyltransferase II deficiency, infantile
267607014	NM_002942.4(ROBO2):c.2834T>C (p.Ile945Thr)	GAGAYTGGAAATTTTGGCCGTGG	Vesicoureteral reflux 2
281865192	NM_025114.3(CEP290):c.2991+1655A>G	GATAYTCACAATTACAACTGGGG, AGATAYTCACAATTACAACTGGG, GAGATAYTCACAATTACAACTGG	Leber congenital amaurosis 10
386833492	NM_000112.3(SLC26A2):c.-26+2T>C	GAGAGGYGAGAAGAGGGAAGCGG	Diastrophic dysplasia
587779773	NM_001101.3(ACTB):c.356T>C (p.Met119Thr)	GAGAAGAYGACCCAGGTGAGTGG	Baraitser-Winter syndrome 1
121913512	NM_000222.2(KIT):c.1924A>G (p.Lys642Glu)	GACTTYGAGTTCAGACATGAGGG, GGACTTYGAGTTCAGACATGAGG	
28939072	NM_006329.3(FBLN5):c.506T>C (p.Ile169Thr)	GACAYTGATGAATGTCGCTATGG	Age-related macular degeneration 3
104894248	NM_000525.3(KCNJ11):c.776A>G (p.His259Arg)	GACAYGGTAGATGATCAGCGGGG, TGACAYGGTAGATGATCAGCGGG, ATGACAYGGTAGATGATCAGCGG	Islet cell hyperplasia
387907132	NM_016464.4(TMEM138):c.287A>G (p.His96Arg)	GACAYGAAGGGAGATGCTGAGGG, AGACAYGAAGGGAGATGCTGAGG	Joubert syndrome 16
121918170	NM_000275.2(OCA2):c.1465A>G (p.Asn489Asp)	GACATYTGGAGGGTCCCCGATGG	Tyrosinase-positive oculocutaneous albinism
122467173	NM_014009.3(FOXP3):c.970T>C (p.Phe324Leu)	GACAGAGYTCCTCCACAACATGG	Insulin-dependent diabetes mellitus secretory diarrhea syndrome
137852268	NM_000133.3(F9):c.1328T>C (p.Ile443Thr)	GAAYATATACCAAGGTATCCCGG	Hereditary factor IX deficiency disease
149054177	NM_001999.3(FBN2):c.3740T>C (p.Met1247Thr)	GAATGTAYGATAATGAACGGAGG	not specified, Macular degeneration, early-onset
137854488	NM_212482.1(FN1):c.2918A>G (p.Tyr973Cys)	GAAGTAAYAGGTGACCCCAGGGG	Glomerulopathy with fibronectin deposits 2
786204027	NM_005957.4(MTHFR):c.1530+2T>C	GAAGGYGTGGTAGGGAGGCACGG, AAGGYGTGGTAGGGAGGCACGGG, AGGYGTGGTAGGGAGGCACGGGG	Homocysteinemia due to MTHFR deficiency
104894223	NM_012193.3(FZD4):c.766A>G (p.Ile256Val)	GAAATAYGATGGGGCGCTCAGGG, AGAAATAYGATGGGGCGCTCAGG	Retinopathy of prematurity
137854474	NM_000138.4(FBN1):c.3793T>C (p.Cys1265Arg)	CTTGYGTTATGATGGATTCATGG	Marfan syndrome
587784418	NM_006306.3(SMC1A):c.3254A>G (p.Tyr1085Cys)	CTTAYAGATCTCATCAATGTTGG	Congenital muscular hypertrophy-cerebral syndrome
81002805	NM_000059.3(BRCA2):c.316+2T>C	CTTAGGYAAGTAATGCAATATGG	Familial cancer of breast, Breast-ovarian cancer, familial 2, Hereditary cancer-predisposing syndrome
121909653	NM_182925.4(FLT4):c.3104A>G (p.His1035Arg)	CTGYGGATGCACTGGGGTGCGGG, TCTGYGGATGCACTGGGGTGCGG	
786205107	NM_031226.2(CYP19A1):c.743+2T>C	CTGTGYAAGTAATACAACTTTGG	Aromatase deficiency
587777037	NM_001283009.1(RTEL1):c.3730T>C (p.Cys1244Arg)	CTGTGTGYGCCAGGGCTGTGGGG	Dyskeratosis congenita, autosomal recessive, 5
794728380	NM_000238.3(KCNH2):c.1945+6T>C	CTGTGAGYGTGCCCAGGGGCGGG, TGAGYGTGCCCAGGGGCGGGCGG	Cardiac arrhythmia
267607987	NM_000251.2(MSH2):c.2005+2T>C	CTGGYAAAAAACCTGGTTTTTGG, TGGYAAAAAACCTGGTTTTTGGG	Hereditary Nonpolyposis Colorectal Neoplasms
397509397	NM_006876.2(B4GAT1):c.1168A>G (p.Asn390Asp)	TGATYTTCAGCCTCCTTTTGGGG, CTGATYTTCAGCCTCCTTTTGGG, GCTGATYTTCAGCCTCCTTTTGG	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A13
121918381	NM_000040.1(APOC3):c.280A>G (p.Thr94Ala)	CTGAAGYTGGTCTGACCTCAGGG, GCTGAAGYTGGTCTGACCTCAGG	
104894919	NM_001015877.1(PHF6):c.769A>G (p.Arg257Gly)	CTCYTGATGTTGTTGTGAGCTGG	Borjeson-Forssman-Lehmann syndrome
267606869	NM_005144.4(HR):c.-218A>G	CTCYAGGGCCGCAGGTTGGAGGG, GCTCYAGGGCCGCAGGTTGGAGG, GGCGCTCYAGGGCCGCAGGTTGG	Marie Unna hereditary hypotrichosis 1
139732572	NM_000146.3(FTL):c.1A>G (p.Met1Val)	CTCAYGGTTGGTTGGCAAGAAGG	L-ferritin deficiency
397515418	NM_018486.2(HDAC8):c.1001A>G (p.His334Arg)	CTCAYGATCTGGGATCTCAGAGG	Cornelia de Lange syndrome 5
372395294	NM_198056.2(SCN5A):c.1247A>G (p.Tyr416Cys)	CTCAYAGGCCATTGCGACCACGG	not provided
104895304	NM_000431.3(MVK):c.803T>C (p.Ile268Thr)	CTCAAYAGATGCCATCTCCCTGG	Hyperimmunoglobulin D with periodic fever, Mevalonic aciduria
587777188	NM_001165899.1(PDE4D):c.1850T>C (p.Ile617Thr)	CTATAYTGTTCATCCCCTCTGGG, ACTATAYTGTTCATCCCCTCTGG	Acrodysostosis 2, with or without hormone resistance
398123026	NM_003867.3(FGF17):c.560A>G (p.Asn187Ser)	CGTGGYTGGGGAAGGGCAGCTGG	Hypogonadotropic hypogonadism 20 with or without anosmia

121964924	NM_001385.2(DPYS):c.1078T>C (p.Trp360Arg)	CGTAATAYGGGAAAAAGGCGTGG, AATAYGGGAAAAAGGCGTGGTGG, ATAYGGGAAAAAGGCGTGGTGGG	Dihydropyrimidinase deficiency
587777301	NM_199189.2(MATR3):c.1864A>G (p.Thr622Ala)	CGGYTGAACTCTCAGTCTTCTGG	Myopathy, distal, 2
200238879	NM_000527.4(LDLR):c.694+2T>C	ACTGCGGYATGGGCGGGGCCAGG, CTGCGGYATGGGCGGGGCCAGGG, CGGYATGGGCGGGGCCAGGGTGG	Familial hypercholesterolemia
142951029	NM_145046.4(CALR3):c.245A>G (p.Lys82Arg)	CGGTYTGAAGCGTGCAGAGATGG	Arrhythmogenic right ventricular cardiomyopathy, Familial hypertrophic cardiomyopathy 19, Hypertrophic cardiomyopathy
786200953	NM_006785.3(MALT1):c.1019-2A>G	CGCYTTGAAAAAAAAAGAAAGGG, TCGCYTTGAAAAAAAAAGAAAGG	Combined immunodeficiency
120074192	NM_000218.2(KCNQ1):c.418A>G (p.Ser140Gly)	CGCYGAAGATGAGGCAGACCAGG	Atrial fibrillation, familial, 3, Atrial fibrillation
267606887	NM_005957.4(MTHFR):c.971A>G (p.Asn324Ser)	CGCGGYTGAGGGTGTAGAAGTGG	Homocystinuria due to MTHFR deficiency
118192117	NM_000540.2(RYR1):c.1205T>C (p.Met402Thr)	CGCAYGATCCACAGCACCAATGG	Congenital myopathy with fiber type disproportion, Central core disease
199473625	NM_198056.2(SCN5A):c.4978A>G (p.Ile1660Val)	CGAYGTTGAAGAGGGCAGGCAGG, AGCCCGAYGTTGAAGAGGGCAGG	Brugada syndrome
794726865	NM_000921.4(PDE3A):c.1333A>G (p.Thr445Ala)	CGAGGYGGTGGTGGTCCAAGTGG	Brachydactyly with hypertension
606231254	NM_005740.2(DNAL4):c.153+2T>C	CGAGGYATTGCCAGCAGTGCAGG	Mirror movements 3
786204826	NM_004771.3(MMP20):c.611A>G (p.His204Arg)	CGAAAYGTGTATCTCCTCCCAGG	Amelogenesis imperfecta, hypomaturation type, IIA2
796053139	NM_021007.2(SCN2A):c.4308+2T>C	CGAAATGYAAGTCTAGTTAGAGG, GAAATGYAAGTCTAGTTAGAGGG	not provided
137854494	NM_005502.3(ABCA1):c.4429T>C (p.Cys1477Arg)	CCTGTGYGTCCCCCAGGGGCAGG, CTGTGYGTCCCCCAGGGGCAGGG, TGTGYGTCCCCCAGGGGCAGGGG, GTGYGTCCCCCAGGGGCAGGGGG	Tangier disease
786205144	NM_001103.3(ACTN2):c.683T>C (p.Met228Thr)	CCTAAAAYGTTGGATGCTGAAGG	Dilated cardiomyopathy 1AA
199919568	NM_007254.3(PNKP):c.1029+2T>C	CCGGYGAGGCCCTGGGGCGGGGG, TCCGGYGAGGCCCTGGGGCGGGG, ATCCGGYGAGGCCCTGGGGCGGG, GATCCGGYGAGGCCCTGGGGCGG	not provided
28939079	NM_018965.3(TREM2):c.401A>G (p.Asp134Gly)	TGAYCCAGGGGGTCTATGGGAGG, CGGTGAYCCAGGGGGTCTATGGG, CCGGTGAYCCAGGGGGTCTATGG	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
193302855	NM_032520.4(GNPTG):c.610-2A>G	CCCYGAAGGTGGAGGATGCAGGG, GCCCYGAAGGTGGAGGATGCAGG	Mucolipidosis III Gamma
111033708	NM_000155.3(GALT):c.499T>C (p.Trp167Arg)	CCCTYGGGTGCAGGTTTGTGAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
28933378	NM_000174.4(GP9):c.70T>C (p.Cys24Arg)	CCCAYGTACCTGCCGCGCCCTGG	Bernard Soulier syndrome, Bernard-Soulier syndrome type C
364897	NM_000157.3(GBA):c.680A>G (p.Asn227Ser)	CCAYTGGTCTTGAGCCAAGTGGG, TCCAYTGGTCTTGAGCCAAGTGG	Gaucher disease, Subacute neuronopathic Gaucher disease, Gaucher disease, type 1
796052551	NM_000833.4(GRIN2A):c.2449A>G (p.Met817Val)	CCAYGTTGTCAATGTCCAGCTGG	not provided
63751006	NM_002087.3(GRN):c.2T>C (p.Met1Thr)	CCAYGTGGACCCTGGTGAGCTGG	Frontotemporal dementia, ubiquitin-positive
786203997	NM_001031.4(RPS28):c.1A>G (p.Met1Val)	TGTCCAYGATGGCGGCGCGGCGG, CCAYGATGGCGGCGCGGCGGCGG	Diamond-Blackfan anemia with microtia and cleft palate
121908595	NM_002755.3(MAP2K1):c.389A>G (p.Tyr130Cys)	CCAYAGAAGCCCACGATGTACGG	Cardiofaciocutaneous syndrome 3, Rasopathy
398122910	NM_000431.3(MVK):c.1039+2T>C	CCAGGYATCCCGGGGGTAGGTGG, CAGGYATCCCGGGGGTAGGTGGG	Porokeratosis, disseminated superficial actinic 1
119474039	NM_020365.4(EIF2B3):c.1037T>C (p.Ile346Thr)	CCAGAYTGTCAGCAAACACCTGG	Leukoencephalopathy with vanishing white matter
587777866	NM_000076.2(CDKN1C):c.*5+2T>C	CCAAGYGAGTACAGCGCACCTGG, CAAGYGAGTACAGCGCACCTGGG, AAGYGAGTACAGCGCACCTGGGG	Beckwith-Wiedemann syndrome
121918530	NM_005587.2(MEF2A):c.788A>G (p.Asn263Ser)	AGAYTACCACCACCTGGTGGAGG, CCAAGAYTACCACCACCTGGTGG	
483352818	NM_000211.4(ITGB2):c.1877+2T>C	CATGYGAGTGCAGGCGGAGCAGG	Leukocyte adhesion deficiency type 1
460184	NM_000186.3(CFH):c.3590T>C (p.Val1197Ala)	CAGYTGAATTTGTGTGTAAACGG	Atypical hemolytic-uremic syndrome 1
121908423	NM_004795.3(KL):c.578A>G (p.His193Arg)	CAGYGGTACAGGGTGACCACGGG, CCAGYGGTACAGGGTGACCACGG	
281860300	NM_005247.2(FGF3):c.146A>G (p.Tyr49Cys)	CAGYAGAGCTTGCGGCGCCGGGG, GCAGYAGAGCTTGCGGCGCCGGG, CGCAGYAGAGCTTGCGGCGCCGG	Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
28935488	NM_000169.2(GLA):c.806T>C (p.Val269Ala)	CAGTTAGYGATTGGCAACTTTGG	Fabry disease

587776514	NM_173560.3(RFX6):c.380+2T>C	CAGTGGYGAGACTCGCCCGCAGG, AGTGGYGAGACTCGCCCGCAGGG	Mitchell-Riley syndrome
104894117	NM_178138.4(LHX3):c.332A>G (p.Tyr111Cys)	CAGGTGGYACACGAAGTCCTGGG	Pituitary hormone deficiency, combined 3
34878913	NM_000184.2(HBG2):c.125T>C (p.Phe42Ser)	CAGAGGTYCTTTGACAGCTTTGG	Cyanosis, transient neonatal
120074124	NM_000543.4(SMPD1):c.911T>C (p.Leu304Pro)	AGCACYTGTGAGGAAGTTCCTGG, GCACYTGTGAGGAAGTTCCTGGG, CACYTGTGAGGAAGTTCCTGGGG	Sphingomyelin/cholesterol lipidosis, Niemann-Pick disease, type A, Niemann-Pick disease, type B
281860272	NM_005211.3(CSF1R):c.2320-2A>G	CACYGAGGGAAAGCACTGCAGGG, GCACYGAGGGAAAGCACTGCAGG	Hereditary diffuse leukoencephalopathy with spheroids
128624216	NM_000033.3(ABCD1):c.443A>G (p.Asn148Ser)	CACTGYTGACGAAGGTAGCAGGG, GCACTGYTGACGAAGGTAGCAGG	Adrenoleukodystrophy
398124257	NM_012463.3(ATP6V0A2):c.825+2T>C	CACTGYGAGTAAGCTGGAAGTGG	Cutis laxa with osteodystrophy
267606679	NM_004183.3(BEST1):c.704T>C (p.Val235Ala)	CACTGGYGTATACACAGGTGAGG	Vitreoretinochoroidopathy dominant
397514518	NM_000344.3(SMN1):c.388T>C (p.Tyr130His)	CACTGGAYATGGAAATAGAGAGG	Kugelberg-Welander disease
143946794	NM_001946.3(DUSP6):c.566A>G (p.Asn189Ser)	CACTAYTGGGGTCTCGGTCAAGG	Hypogonadotropic hypogonadism 19 with or without anosmia
397516076	NM_000256.3(MYBPC3):c.821+2T>C	GCACGYGAGTGGCCATCCTCAGG, CACGYGAGTGGCCATCCTCAGGG	Familial hypertrophic cardiomyopathy 4, not specified
149977726	NM_001257988.1(TYMP):c.665A>G (p.Lys222Arg)	CACGAGTYTCTTACTGAGAATGG, GAGTYTCTTACTGAGAATGGAGG	
121917770	NM_003361.3(UMOD):c.383A>G (p.Asn128Ser)	CACAYTGACACATGTGGCCAGGG, CCACAYTGACACATGTGGCCAGG	Familial juvenile gout
121909008	NM_000492.3(CFTR):c.2738A>G (p.Tyr913Cys)	CACATAAYACGAACTGGTGCTGG	Cystic fibrosis
137852819	NM_003688.3(CASK):c.2740T>C (p.Trp914Arg)	CACAGYGGGTCCCTGTCTCCTGG, ACAGYGGGTCCCTGTCTCCTGGG	FG syndrome 4
74315320	NM_024009.2(GJB3):c.421A>G (p.Ile141Val)	CAAYGATGAGCTTGAAGATGAGG	Deafness, autosomal recessive
80356747	NM_001701.3(BAAT):c.967A>G (p.Ile323Val)	CAAYGAAGAGGAATTGCCCCTGG	Atypical hemolytic-uremic syndrome 1
180177324	NM_012203.1(GRHPR):c.934A>G (p.Asn312Asp)	CAAGTYGTTAGCTGCCAACAAGG	Primary hyperoxaluria, type II
281860274	NM_005211.3(CSF1R):c.2381T>C (p.Ile794Thr)	CAAGAYTGGGGACTTCGGGCTGG	Hereditary diffuse leukoencephalopathy with spheroids
398122908	NM_005334.2(HCFC1):c.-970T>C	CAAGAYGGCGGCTCCCAGGGAGG	Mental retardation 3, X-linked
548076633	NM_002693.2(POLG):c.3470A>G (p.Asn1157Ser)	CAAGAGGYTGGTGATCTGCAAGG	not provided
120074146	NM_000019.3(ACAT1):c.935T>C (p.Ile312Thr)	CAAGAAYAGTAGGTAAGGCCAGG	Deficiency of acetyl-CoA acetyltransferase
397514489	NM_005340.6(HINT1):c.250T>C (p.Cys84Arg)	CAAGAAAYGTGCTGCTGATCTGG, AAGAAAYGTGCTGCTGATCTGGG	Gamstorp-Wohlfart syndrome
587783539	NM_178151.2(DCX):c.2T>C (p.Met1Thr)	CAAAATAYGGAACTTGATTTTGG	Heterotopia
104894765	NM_005448.2(BMP15):c.704A>G (p.Tyr235Cys)	ATTGAAAYAGAGTAACAAGAAGG	Ovarian dysgenesis 2
137852429	NM_000132.3(F8):c.1892A>G (p.Asn631Ser)	ATGYTGGAGGCTTGGAACTCTGG	Hereditary factor VIII deficiency disease
72558441	NM_000531.5(OTC):c.779T>C (p.Leu260Ser)	ATGTATYAATTACAGACACTTGG	not provided
398123765	NM_003494.3(DYSF):c.1284+2T>C	ATGGYAAGGAGCAAGGGAGCAGG	Limb-girdle muscular dystrophy, type 2B
387906924	NM_020191.2(MRPS22):c.644T>C (p.Leu215Pro)	ATCYTAGGGTAAGGTGACTTAGG	Combined oxidative phosphorylation deficiency 5
397518039	NM_206933.2(USH2A):c.8559-2A>G	ATCYAAAGCAAAAGACAAGCAGG	Retinitis pigmentosa, Usher syndrome, type 2A
5742905	NM_000071.2(CBS):c.833T>C (p.Ile278Thr)	ATCAYTGGGGTGGATCCCGAAGG, TCAYTGGGGTGGATCCCGAAGGG	Homocystinuria due to CBS deficiency, Homocystinuria, pyridoxine-responsive
397507473	NM_004333.4(BRAF):c.1403T>C (p.Phe468Ser)	ATCATYTGGAACAGTCTACAAGG, TCATYTGGAACAGTCTACAAGGG	Cardiofaciocutaneous syndrome, Rasopathy
786204056	NM_000264.3(PTCH1):c.3168+2T>C	ATCATTGYGAGTGTATTATAAGG, TCATTGYGAGTGTATTATAAGGG, CATTGYGAGTGTATTATAAGGGG	Gorlin syndrome
72558484	NM_000531.5(OTC):c.1005+2T>C	ATCATGGYAAGCAAGAAACAAGG	not provided
199473074	NM_000335.4(SCN5A):c.688A>G (p.Ile230Val)	ATAYAGTTTTCAGGGCCCGGAGG, CTGATAYAGTTTTCAGGGCCCGG	Brugada syndrome
111033273	NM_206933.2(USH2A):c.1606T>C (p.Cys536Arg)	ATATAGAYGCCTCTGCTCCCAGG	Usher syndrome, type 2A
72556290	NM_000531.5(OTC):c.542A>G (p.Glu181Gly)	ATAGTGTYCCTAAAAGGCACGGG	not provided
121918711	NM_004612.3(TGFBR1):c.1199A>G (p.Asp400Gly)	ATAGATGYCAGCACGTTTGAAGG	Loeys-Dietz syndrome 1
104886288	NM_000495.4(COL4A5):c.4699T>C (p.Cys1567Arg)	AGTAYGTGAAGCTCCAGCTGTGG	Alport syndrome, X-linked recessive

144637717	NM_016725.2(FOLR1):c.493+2T>C	CTTCAGGYGAGGGCTGGGGTGGG, AGGYGAGGGCTGGGGTGGGCAGG	not provided
72558492	NM_000531.5(OTC):c.1034A>G (p.Tyr345Cys)	AGGTGAGYAATCTGTCAGCAGGG	not provided
62638745	NM_000121.3(EPOR):c.1460A>G (p.Asn487Ser)	AGGGYTGGAGTAGGGGCCATCGG	Acute myeloid leukemia, M6 type, Familial erythrocytosis, 1
387907021	NM_031427.3(DNAL1):c.449A>G (p.Asn150Ser)	AGGGAYTGCCTACAAACACCAGG	Kartagener syndrome, Ciliary dyskinesia, primary, 16
397514488	NM_001161581.1(POC1A):c.398T>C (p.Leu133Pro)	AGCYGTGGGACAAGAGCAGCCGG	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
154774633	NM_017882.2(CLN6):c.200T>C (p.Leu67Pro)	AGCYGGTATTCCCTCTCGAGTGG	Adult neuronal ceroid lipofuscinosis
111033700	NM_000155.3(GALT):c.482T>C (p.Leu161Pro)	AGCYGGGTGCCCAGTACCCTTGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
128621198	NM_000061.2(BTK):c.1223T>C (p.Leu408Pro)	GAGCYGGGGACTGGACAATTTGG, AGCYGGGGACTGGACAATTTGGG	X-linked agammaglobulinemia
137852611	NM_000211.4(ITGB2):c.446T>C (p.Leu149Pro)	AGCYAGGTGGCGACCTGCTCCGG	Leukocyte adhesion deficiency
121908838	NM_003722.4(TP63):c.697A>G (p.Lys233Glu)	AGCTTYTTTGTAGACAGGCATGG	Split-hand/foot malformation 4
397515869	NM_000169.2(GLA):c.1153A>G (p.Thr385Ala)	AGCTGTGYGATGAAGCAGGCAGG	not specified
118204064	NM_000237.2(LPL):c.548A>G (p.Asp183Gly)	GCTGGAYCGAGGCCTTAAAAGGG, AGCTGGAYCGAGGCCTTAAAAGG	Hyperlipoproteinemia, type I
128620186	NM_000061.2(BTK):c.2T>C (p.Met1Thr)	AGCTAYGGCCGCAGTGATTCTGG	X-linked agammaglobulinemia
786204132	NM_014946.3(SPAST):c.1165A>G (p.Thr389Ala)	ATTGYCTTCCCATTCCCAGGTGG, AGCATTGYCTTCCCATTCCCAGG	Spastic paraplegia 4, autosomal dominant
199473661	NM_000218.2(KCNQ1):c.550T>C (p.Tyr184His)	CAGCAAGBACGTGGGCCTCTGGG, AGCAAGBACGTGGGCCTCTGGGG, GCAAGBACGTGGGCCTCTGGGGG	Congenital long QT syndrome, Cardiac arrhythmia
387907129	NM_024599.5(RHBDF2):c.557T>C (p.Ile186Thr)	AGAYTGTGGATCCGCTGGCCCGG	Howel-Evans syndrome
387906702	NM_006306.3(SMC1A):c.2351T>C (p.Ile784Thr)	AGAYTGGTGTGCGCAACATCCGG	Congenital muscular hypertrophy-cerebral syndrome
193929348	NM_000525.3(KCNJ11):c.544A>G (p.Ile182Val)	AGAYGAGGGTCTCAGCCCTGCGG	Permanent neonatal diabetes mellitus
121908934	NM_004086.2(COCH):c.1535T>C (p.Met512Thr)	AGATAYGGCTTCTAAACCGAAGG	Deafness, autosomal dominant 9
397514377	NM_000060.3(BTD):c.641A>G (p.Asn214Ser)	AGAGGYTGTGTTTACGGTAGCGG	Biotinidase deficiency
72552295	NM_000531.5(OTC):c.2T>C (p.Met1Thr)	AGAAGAYGCTGTTTAATCTGAGG	not provided
201893545	NM_016247.3(IMPG2):c.370T>C (p.Phe124Leu)	ACTYTTTGGGATCGACTTCCTGG	Macular dystrophy, vitelliform, 5
121434469	m.4290T>C	ACTYTGATAGAGTAAATAATAGG	
121918733	NM_006920.4(SCN1A):c.269T>C (p.Phe90Ser)	ACTTYTATAGTATTGAATAAAGG, CTTYTATAGTATTGAATAAAGGG	Severe myoclonic epilepsy in infancy
121434471	m.4291T>C	ACTTYGATAGAGTAAATAATAGG	Hypertension, hypercholesterolemia, and hypomagnesemia, mitochondrial
606231289	NM_001302946.1(TRNT1):c.497T>C (p.Leu166Ser)	ACTTYATTTGACTACTTTAATGG	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
63750067	NM_000517.4(HBA2):c.*92A>G	CTTYATTCAAAGACCAGGAAGGG, ACTTYATTCAAAGACCAGGAAGG	Hemoglobin H disease, nondeletional
121918734	NM_006920.4(SCN1A):c.272T>C (p.Ile91Thr)	ACTTTTAYAGTATTGAATAAAGG, CTTTTAYAGTATTGAATAAAGGG	Severe myoclonic epilepsy in infancy
137854557	NM_000267.3(NF1):c.1466A>G (p.Tyr489Cys)	ACTTAYAGCTTCTTGTCTCCAGG	Neurofibromatosis, type 1
397514626	NM_018344.5(SLC29A3):c.607T>C (p.Ser203Pro)	ACTGATAYCAGGTGAGAGCCAGG, CTGATAYCAGGTGAGAGCCAGGG	Histiocytosis-lymphadenopathy plus syndrome
118204440	NM_000512.4(GALNS):c.1460A>G (p.Asn487Ser)	ACGYTGAGCTGGGGCTGCGCGGG, CACGYTGAGCTGGGGCTGCGCGG	Mucopolysaccharidosis, MPS-IV-A
587776843	NG_012088.1:g.2209A>G	ACCYTATGATCCGCCCGCCTTGG	
137853033	NM_001080463.1(DYNC2H1):c.4610A>G (p.Gln1537Arg)	ACCYGTGAAGGGAACAGAGATGG	Short-rib thoracic dysplasia 3 with or without polydactyly
28933698	NM_000435.2(NOTCH3):c.1363T>C (p.Cys455Arg)	TTCACCYGTATCTGTATGGCAGG, ACCYGTATCTGTATGGCAGGTGG	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
587776766	NM_000463.2(UGT1A1):c.1085-2A>G	ACCYGAGATGCAAAATAGGGAGG, GTGACCYGAGATGCAAAATAGGG, GGTGACCYGAGATGCAAAATAGG	Crigler Najjar syndrome, type 1
587781628	NM_001128425.1(MUTYH):c.1187-2A>G	ACCYGAGAGGGAGGGCAGCCAGG	Hereditary cancer-predisposing syndrome, Carcinoma of colon
61755817	NM_000322.4(PRPH2):c.736T>C (p.Trp246Arg)	ACCTGYGGGTGCGTGGCTGCAGG, CCTGYGGGTGCGTGGCTGCAGGG	Retinitis pigmentosa
121909184	NM_001089.2(ABCA3):c.1702A>G (p.Asn568Asp)	ACCGTYGTGGCCCAGCAGGACGG	Surfactant metabolism dysfunction, pulmonary, 3

121434466	m.4269A>G	ACAYATTTCTTAGGTTTGAGGGG, GACAYATTTCTTAGGTTTGAGGG, AGACAYATTTCTTAGGTTTGAGG	
794726768	NM_001165963.1(SCN1A):c.1048A>G (p.Met350Val)	ACAYATATCCCTCTGGACATTGG	Severe myoclonic epilepsy in infancy
28934876	NM_001382.3(DPAGT1):c.509A>G (p.Tyr170Cys)	ACAYAGTACAGGATTCCTGCGGG, GACAYAGTACAGGATTCCTGCGG	Congenital disorder of glycosylation type 1J
104894749	NM_000054.4(AVPR2):c.614A>G (p.Tyr205Cys)	ACAYAGGTGCGACGGCCCCAGGG, GACAYAGGTGCGACGGCCCCAGG	Nephrogenic diabetes insipidus, Nephrogenic diabetes insipidus, X-linked
128621205	NM_000061.2(BTK):c.1741T>C (p.Trp581Arg)	ACATTYGGGCTTTTGGTAAGTGG	X-linked agammaglobulinemia
28940892	NM_000529.2(MC2R):c.761A>G (p.Tyr254Cys)	ACATGYAGCAGGCGCAGTAGGGG, GACATGYAGCAGGCGCAGTAGGG, AGACATGYAGCAGGCGCAGTAGG	ACTH resistance
794726844	NM_001165963.1(SCN1A):c.1046A>G (p.Tyr349Cys)	ACATAYATCCCTCTGGACATTGG	Severe myoclonic epilepsy in infancy
587783083	NM_003159.2(CDKL5):c.449A>G (p.Lys150Arg)	ACAGTYTTAGGACATCATTGTGG	not provided
397514651	NM_000108.4(DLD):c.140T>C (p.Ile47Thr)	ACAGTTAYAGGTTCTGGTCCTGG, GTTAYAGGTTCTGGTCCTGGAGG	Maple syrup urine disease, type 3
794727060	NM_001848.2(COL6A1):c.957+2T>C	ACAAGGYGAGCGTGGGCTGCTGG, CAAGGYGAGCGTGGGCTGCTGGG	Ullrich congenital muscular dystrophy, Bethlem myopathy
72554346	NM_000531.5(OTC):c.284T>C (p.Leu95Ser)	ACAAGATYGTCTACAGAAACAGG	not provided
483353031	NM_002136.2(HNRNPA1):c.841T>C (p.Phe281Leu)	AATYTTGGAGGCAGAAGCTCTGG	Chronic progressive multiple sclerosis
104894271	NM_000315.2(PTH):c.52T>C (p.Cys18Arg)	AATTYGTTTTCTTACAAAATCGG	Hypoparathyroidism familial isolated
267608260	NM_015599.2(PGM3):c.248T>C (p.Leu83Ser)	AATGTYGGCACCATCCTGGGAGG	Immunodeficiency 23
267606900	NM_018109.3(MTPAP):c.1432A>G (p.Asn478Asp)	AATGGATYCTGAATGTACAGAGG	Ataxia, spastic, 4, autosomal recessive
796053169	NM_021007.2(SCN2A):c.387-2A>G	AATAAAGYAGAATATCGTCAAGG	not provided
104894937	NM_000116.4(TAZ):c.352T>C (p.Cys118Arg)	AAGYGTGTGCCTGTGTGCCGAGG	3-Methylglutaconic aciduria type 2
104893911	NM_001018077.1(NR3C1):c.1712T>C (p.Val571Ala)	AAGYGATTGCAGCAGTGAAATGG	Pseudohermaphroditism, female, with hypokalemia, due to glucocorticoid resistance
397514472	NM_004813.2(PEX16):c.992A>G (p.Tyr331Cys)	AAGYAGATTTTCTGCCAGGTGGG, GAAGYAGATTTTCTGCCAGGTGG, GTAGAAGYAGATTTTCTGCCAGG	Peroxisome biogenesis disorder 8B
121918407	NM_001083112.2(GPD2):c.1904T>C (p.Phe635Ser)	AAGTYTGATGCAGACCAGAAAGG	Diabetes mellitus type 2
63751110	NM_000251.2(MSH2):c.595T>C (p.Cys199Arg)	AAGGAAYGTGTTTTACCCGGAGG	Hereditary Nonpolyposis Colorectal Neoplasms
119450945	NM_000026.2(ADSL):c.674T>C (p.Met225Thr)	AAGAYGGTGACAGAAAAGGCAGG	Adenylosuccinate lyase deficiency
113993988	NM_002863.4(PYGL):c.2461T>C (p.Tyr821His)	AAGAAYATGCCCAAAACATCTGG	Glycogen storage disease, type VI
119485091	NM_022041.3(GAN):c.1268T>C (p.Ile423Thr)	AAGAAAAYCTACGCCATGGGTGG, AAAAYCTACGCCATGGGTGGAGG	Giant axonal neuropathy
137852419	NM_000132.3(F8):c.1660A>G (p.Ser554Gly)	AACYAGAGTAATAGCGGGTCAGG	Hereditary factor VIII deficiency disease
121964967	NM_000071.2(CBS):c.1150A>G (p.Lys384Glu)	AACTYGGTCCTGCGGGATGGGGG, GAACTYGGTCCTGCGGGATGGGG, GGAACTYGGTCCTGCGGGATGGG, AGGAACTYGGTCCTGCGGGATGG	Homocystinuria, pyridoxine-responsive
137852376	NM_000132.3(F8):c.1754T>C (p.Ile585Thr)	AACAGAYAATGTCAGACAAGAGG	Hereditary factor VIII deficiency disease
121917930	NM_006920.4(SCN1A):c.3577T>C (p.Trp1193Arg)	AACAAYGGTGGAACCTGAGAAGG	Generalized epilepsy with febrile seizures plus, type 1, Generalized epilepsy with febrile seizures plus, type 2
28939717	NM_003907.2(EIF2B5):c.271A>G (p.Thr91Ala)	AAATGYTTCCTGTACACCTGTGG	Leukoencephalopathy with vanishing white matter
80357276	NM_007294.3(BRCA1):c.122A>G (p.His41Arg)	AAATATGYGGTCACACTTTGTGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
397515897	NM_000256.3(MYBPC3):c.1351+2T>C	AAAGGYGGGCCTGGGACCTGAGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397514491	NM_005340.6(HINT1):c.152A>G (p.His51Arg)	AAAAYGTGTTGGTGCTTGAGGGG, GAAAAYGTGTTGGTGCTTGAGGG, AGAAAAYGTGTTGGTGCTTGAGG	Gamstorp-Wohlfart syndrome
387907164	NM_020894.2(UVSSA):c.94T>C (p.Cys32Arg)	AAAATTYGCAAGTATGTCTTAGG, AAATTYGCAAGTATGTCTTAGGG	UV-sensitive syndrome 3
118161496	NM_025152.2(NUBPL):c.815-27T>C	TGGTTCYAATGGATGTCTGCTGG, GGTTCYAATGGATGTCTGCTGGG	Mitochondrial complex I deficiency
764313717	NM_005609.2(PYGM):c.425_528del	TGGCTGYCAGGGACCCAGCAAGG, CTGYCAGGGACCCAGCAAGGAGG	
28934568	NM_003242.5(TGFBR2):c.923T>C (p.Leu308Pro)	AGTTCCYGACGGCTGAGGAGCGG	Loeys-Dietz syndrome 2

121913461	NM_007313.2(ABL1):c.814T>C (p.Tyr272His)	CCAGYACGGGGAGGTGTACGAGG, CAGYACGGGGAGGTGTACGAGGG	
377750405	NM_173551.4(ANKS6):c.1322A>G (p.Gln441Arg)	AGGGCYGTCGGACCTTCGAGTGG, GGGCYGTCGGACCTTCGAGTGGG, GGCYGTCGGACCTTCGAGTGGGG	Nephronophthisis 16
57639980	NM_001927.3(DES):c.1034T>C (p.Leu345Pro)	ATTCCCYGATGAGGCAGATGCGG, TTCCCYGATGAGGCAGATGCGGG	Myofibrillar myopathy 1
147391618	NM_020320.3(RARS2):c.35A>G (p.Gln12Arg)	ATACCYGGCAAGCAATAGCGCGG	Pontocerebellar hypoplasia type 6
182650126	NM_002977.3(SCN9A):c.2215A>G (p.Ile739Val)	GTAAYTGCAAGATCTACAAAAGG	Small fiber neuropathy
80358278	NM_004700.3(KCNQ4):c.842T>C (p.Leu281Ser)	ACATYGACAACCATCGGCTATGG	DFNA 2 Nonsyndromic Hearing Loss
786204012	NM_005957.4(MTHFR):c.388T>C (p.Cys130Arg)	GACCYGCTGCCGTCAGCGCCTGG	Homocysteinemia due to MTHFR deficiency
786204037	NM_005957.4(MTHFR):c.1883T>C (p.Leu628Pro)	TCCCACYGGACAACTGCCTCTGG	Homocysteinemia due to MTHFR deficiency
202147607	NM_000140.3(FECH):c.1137+3A>G	GTAGAYACCTTAGAGAACAATGG	Erythropoietic protoporphyria
122456136	NM_005183.3(CACNA1F):c.2267T>C (p.Ile756Thr)	TGCCAYTGCTGTGGACAACCTGG	
786204851	NM_007374.2(SIX6):c.110T>C (p.Leu37Pro)	GTCGCYGCCCGTGGCCCCTGCGG	Cataract, microphthalmia and nystagmus
794728167	NM_000138.4(FBN1):c.1468+2T>C	ATTGGYACGTGATCCATCCTAGG	Thoracic aortic aneurysms and aortic dissections
121964909	NM_000027.3(AGA):c.214T>C (p.Ser72Pro)	GACGGCYCTGTAGGCTTTGGAGG	Aspartylglycosaminuria
121964978	NM_000170.2(GLDC):c.2T>C (p.Met1Thr)	CGGCCAYGCAGTCCTGTGCCAGG, GGCCAYGCAGTCCTGTGCCAGGG	Non-ketotic hyperglycinemia
121965008	NM_000398.6(CYB5R3):c.446T>C (p.Leu149Pro)	CTGCYGGTCTACCAGGGCAAAGG	METHEMOGLOBINEMIA, TYPE I
121965064	NM_000128.3(F11):c.901T>C (p.Phe301Leu)	TGATYTCTTGGGAGAAGAACTGG	Hereditary factor XI deficiency disease
45517398	NM_000548.3(TSC2):c.5150T>C (p.Leu1717Pro)	GCCCYGCACGCAAATGTGAGTGG, CCCYGCACGCAAATGTGAGTGGG	Tuberous sclerosis syndrome
786205857	NM_015662.2(IFT172):c.770T>C (p.Leu257Pro)	TTGTGCYAGGAAGTTATGACAGG	RETINITIS PIGMENTOSA 71
786205904	NM_001135669.1(XPR1):c.653T>C (p.Leu218Ser)	GCGTTYACGTGTCCCCCCTTTGG, CGTTYACGTGTCCCCCCTTTGGG	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 6
104893704	NM_000388.3(CASR):c.2641T>C (p.Phe881Leu)	ACGCTYTCAAGGTGGCTGCCCGG, CGCTYTCAAGGTGGCTGCCCGGG	Hypercalciuric hypercalcemia
104893747	NM_198159.2(MITF):c.1195T>C (p.Ser399Pro)	ACTTYCCCTTATTCCATCCACGG, CTTYCCCTTATTCCATCCACGGG	Waardenburg syndrome type 2A
104893770	NM_000539.3(RHO):c.133T>C (p.Phe45Leu)	CATGYTTCTGCTGATCGTGCTGG, ATGYTTCTGCTGATCGTGCTGGG	Retinitis pigmentosa 4
28937596	NM_003907.2(EIF2B5):c.1882T>C (p.Trp628Arg)	AGGCCYGGAGCCCTGTTTTTAGG	Leukoencephalopathy with vanishing white matter
104893876	NM_001151.3(SLC25A4):c.293T>C (p.Leu98Pro)	GCAGCYCTTCTTAGGGGGTGTGG	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 2
104893883	NM_006005.3(WFS1):c.2486T>C (p.Leu829Pro)	ACCATCCYGGAGGGCCGCCTGGG	WFS1-Related Disorders
104893962	NM_000165.4(GJA1):c.52T>C (p.Ser18Pro)	CTACYCAACTGCTGGAGGGAAGG	Oculodentodigital dysplasia
104893978	NM_000434.3(NEU1):c.718T>C (p.Trp240Arg)	GCCTCCYGGCGCTACGGAAGTGG, CCTCCYGGCGCTACGGAAGTGGG, CTCCYGGCGCTACGGAAGTGGGG	Sialidosis, type II
104894092	NM_002546.3(TNFRSF11B):c.349T>C (p.Phe117Leu)	TAGAGYTCTGCTTGAAACATAGG	Hyperphosphatasemia with bone disease
104894135	NM_000102.3(CYP17A1):c.316T>C (p.Ser106Pro)	CATCGCGYCCAACAACCGTAAGG, ATCGCGYCCAACAACCGTAAGGG	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
104894151	NM_000102.3(CYP17A1):c.1358T>C (p.Phe453Ser)	AGCTCTYCCTCATCATGGCCTGG	Combined partial 17-alpha-hydroxylase/17,20-lyase deficiency
36015961	NM_000518.4(HBB):c.344T>C (p.Leu115Pro)	TGTGTGCYGGCCCATCACTTTGG	Beta thalassemia intermedia
104894472	NM_152443.2(RDH12):c.523T>C (p.Ser175Pro)	TCCYCGGTGGCTCACCACATTGG	Leber congenital amaurosis 13
104894587	NM_004870.3(MPDU1):c.356T>C (p.Leu119Pro)	TTCCYGGTCATGCACTACAGAGG	Congenital disorder of glycosylation type 1F
104894588	NM_004870.3(MPDU1):c.2T>C (p.Met1Thr)	AATAYGGCGGCCGAGGCGGACGG	Congenital disorder of glycosylation type 1F
104894626	NM_000304.3(PMP22):c.82T>C (p.Trp28Arg)	TAGCAAYGGATCGTGGGCAATGG	Charcot-Marie-Tooth disease, type IE
104894631	NM_018129.3(PNPO):c.784T>C (p.Ter262Gln)	ACCTYAACTCTGGGACCTGCTGG	"Pyridoxal 5-phosphate-dependent epilepsy"
104894703	NM_032551.4(KISS1R):c.305T>C (p.Leu102Pro)	GCCCTGCYGTACCCGCTGCCCGG, TGCYGTACCCGCTGCCCGGCTGG	
104894826	NM_000166.5(GJB1):c.407T>C (p.Val136Ala)	ATGYCATCAGCGTGGTGTTCCGG	Dejerine-Sottas disease, X-linked hereditary motor and sensory neuropathy

104894859	NM_001122606.1(LAMP2):c.961T>C (p.Trp321Arg)	CAGCTACYGGGATGCCCCCCTGG, AGCTACYGGGATGCCCCCCTGGG	Danon disease
104894931	NM_006517.4(SLC16A2):c.1313T>C (p.Leu438Pro)	TGAGCYGGTGGGCCCAATGCAGG	Allan-Herndon-Dudley syndrome
104894935	NM_000330.3(RS1):c.38T>C (p.Leu13Pro)	TTACTTCYCTTTGGCTATGAAGG	Juvenile retinoschisis
104895217	NM_001065.3(TNFRSF1A):c.175T>C (p.Cys59Arg)	TGCYGTACCAAGTGCCACAAAGG	TNF receptor-associated periodic fever syndrome (TRAPS)
143889283	NM_003793.3(CTSF):c.692A>G (p.Tyr231Cys)	CTCCAYACTGAGCTGTGCCACGG	Ceroid lipofuscinosis, neuronal, 13
122459147	NM_001159702.2(FHL1):c.310T>C (p.Cys104Arg)	GGGGYGCTTCAAGGCCATTGTGG	Myopathy, reducing body, X-linked, childhood-onset
74552543	NM_020184.3(CNNM4):c.971T>C (p.Leu324Pro)	AAGCTCCYGGACTTTTTTCTGGG	Cone-rod dystrophy amelogenesis imperfecta
199476117	m.10158T>C	AAAYCCACCCCTTACGAGTGCGG	Leigh disease, Leigh syndrome due to mitochondrial complex I deficiency, Mitochondrial complex I deficiency
794727808	NM_020451.2(SEPN1):c.872+2T>C	TTCCGGYGAGTGGGCCACACTGG	Congenital myopathy with fiber type disproportion, Eichsfeld type congenital muscular dystrophy
140547520	NM_005022.3(PFN1):c.350A>G (p.Glu117Gly)	CACCTYCTTTGCCCATCAGCAGG	Amyotrophic lateral sclerosis 18
397514359	NM_000060.3(BTD):c.445T>C (p.Phe149Leu)	TCACCGCYTCAATGACACAGAGG	Biotinidase deficiency
207460001	m.15197T>C	CTAYCCGCCATCCCATACATTGG	Exercise intolerance
397514406	NM_000060.3(BTD):c.1214T>C (p.Leu405Pro)	TTCACCCYGGTCCCTGTCTGGGG	Biotinidase deficiency
397514516	NM_006177.3(NRL):c.287T>C (p.Met96Thr)	GAGGCCAYGGAGCTGCTGCAGGG	Retinitis pigmentosa 27
72554312	NM_000531.5(OTC):c.134T>C (p.Leu45Pro)	CTCACTCYAAAAAACTTTACCGG	Ornithine carbamoyltransferase deficiency
397514569	NM_178012.4(TUBB2B):c.350T>C (p.Leu117Pro)	GGTCCYGGATGTGGTGAGGAAGG	Polymicrogyria, asymmetric
397514571	NM_000431.3(MVK):c.122T>C (p.Leu41Pro)	CGGCYTCAACCCCACAGCAATGG, GGCYTCAACCCCACAGCAATGGG	Porokeratosis, disseminated superficial actinic 1
794728390	NM_000238.3(KCNH2):c.2396T>C (p.Leu799Pro)	GCCATCCYGGGTATGGGGTGGGG, CCATCCYGGGTATGGGGTGGGGG, CATCCYGGGTATGGGGTGGGGGG	Cardiac arrhythmia
397514713	NM_001199107.1(TBC1D24):c.686T>C (p.Phe229Ser)	GGTCTYTGACGTCTTCCTGGTGG	Early infantile epileptic encephalopathy 16
397514719	NM_080605.3(B3GALT6):c.193A>G (p.Ser65Gly)	CGCYGGCCACCAGCACTGCCAGG	Spondyloepimetaphyseal dysplasia with joint laxity
730880608	NM_000256.3(MYBPC3):c.3796T>C (p.Cys1266Arg)	GAGYGCCGCCTGGAGGTGCGAGG	Cardiomyopathy
397515329	NM_001382.3(DPAGT1):c.503T>C (p.Leu168Pro)	AATCCYGTACTATGTCTACATGG, ATCCYGTACTATGTCTACATGGG, TCCYGTACTATGTCTACATGGGG	Congenital disorder of glycosylation type 1J
397515465	NM_018127.6(ELAC2):c.460T>C (p.Phe154Leu)	ATAYTTTCTGGTCCATTGAAAGG	Combined oxidative phosphorylation deficiency 17
397515557	NM_005211.3(CSF1R):c.2483T>C (p.Phe828Ser)	CATCTYTGACTGTGTCTACACGG	Hereditary diffuse leukoencephalopathy with spheroids
397515599	NM_194248.2(OTOF):c.3413T>C (p.Leu1138Pro)	AGGTGCYGTTCTGGGGCCTACGG, GGTGCYGTTCTGGGGCCTACGGG	Deafness, autosomal recessive 9
397515766	NM_000138.4(FBN1):c.2341T>C (p.Cys781Arg)	GGACAAYGTAGAAATACTCCTGG	Marfan syndrome
565779970	NM_001429.3(EP300):c.3573T>A (p.Tyr1191Ter)	CTTAYTACAGTTACCAGAACAGG	Rubinstein-Taybi syndrome 2
786200938	NM_080605.3(B3GALT6):c.1A>G (p.Met1Val)	AGCTTCAYGGCGCCCGCGCCGGG, TCAYGGCGCCCGCGCCGGGCCGG	Spondyloepimetaphyseal dysplasia with joint laxity
28942087	NM_000229.1(LCAT):c.698T>C (p.Leu233Pro)	ATCTCTCYTGGGGCTCCCTGGGG, TCTCYTGGGGCTCCCTGGGGTGG	Norum disease
128621203	NM_000061.2(BTK):c.1625T>C (p.Leu542Pro)	TCGGCCYGTCCAGGTGAGTGTGG	X-linked agammaglobulinemia with growth hormone deficiency
397515412	NM_006383.3(CIB2):c.368T>C (p.Ile123Thr)	CTTCAYCTGCAAGGAGGACCTGG	Deafness, autosomal recessive 48
193929364	NM_000352.4(ABCC8):c.404T>C (p.Leu135Pro)	AAGCYGCTAATTGGTAGGTGAGG	Permanent neonatal diabetes mellitus
730880872	NM_000257.3(MYH7):c.1400T>C (p.Ile467Thr)	TCGAGAYCTTCGATGTGAGTTGG, CGAGAYCTTCGATGTGAGTTGGG	Cardiomyopathy
80356474	NM_002977.3(SCN9A):c.2543T>C (p.Ile848Thr)	AAGATCAYTGGTAACTCAGTAGG, AGATCAYTGGTAACTCAGTAGGG, GATCAYTGGTAACTCAGTAGGGG	Primary erythromelalgia
80356489	NM_001164277.1(SLC37A4):c.352T>C (p.Trp118Arg)	GGGCYGGCCCCCATGTGGGAAGG	Glucose-6-phosphate transport defect
80356536	NM_152296.4(ATP1A3):c.2338T>C (p.Phe780Leu)	GCCCYTCCTGCTGTTCATCATGG	Dystonia 12
80356596	NM_194248.2(OTOF):c.3032T>C (p.Leu1011Pro)	GATGCYGGTGTTCGACAACCTGG	Deafness, autosomal recessive 9, Auditory neuropathy, autosomal recessive, 1

80356689	NM_000083.2(CLCN1):c.857T>C (p.Val286Ala)	AGGAGYGCTATTTAGCATCGAGG	Myotonia congenita
118203884	m.4409T>C	AGGYCAGCTAAATAAGCTATCGG	Mitochondrial myopathy
587777625	NM_173596.2(SLC39A5):c.911T>C (p.Met304Thr)	AGAACAYGCTGGGGCTTTTGCGG	Myopia 24, autosomal dominant
587783087	NM_003159.2(CDKL5):c.602T>C (p.Leu201Pro)	ATTCYTGGGGAGCTTAGCGATGG	not provided
118203951	NM_013319.2(UBIAD1):c.511T>C (p.Ser171Pro)	TCTGGCYCCTTTCTCTACACAGG, GGCYCCTTTCTCTACACAGGAGG	Schnyder crystalline corneal dystrophy
118204017	NM_000018.3(ACADVL):c.1372T>C (p.Phe458Leu)	TCGCATCYTCCGGATCTTTGAGG, CGCATCYTCCGGATCTTTGAGGG, GCATCYTCCGGATCTTTGAGGGG	Very long chain acyl-CoA dehydrogenase deficiency
397518466	NM_000833.4(GRIN2A):c.2T>C (p.Met1Thr)	CTAYGGGCAGAGTGGGCTATTGG	Focal epilepsy with speech disorder with or without mental retardation
118204069	NM_000237.2(LPL):c.337T>C (p.Trp113Arg)	GGACYGGCTGTCACGGGCTCAGG	Hyperlipoproteinemia, type I
118204080	NM_000237.2(LPL):c.755T>C (p.Ile252Thr)	GTGAYTGCAGAGAGAGGACTTGG	Hyperlipoproteinemia, type I
118204111	NM_000190.3(HMBS):c.739T>C (p.Cys247Arg)	GCTTCGCYGCATCGCTGAAAGGG	Acute intermittent porphyria
80357438	NM_007294.3(BRCA1):c.65T>C (p.Leu22Ser)	AAATCTYAGAGTGTCCCATCTGG	Familial cancer of breast, Breast-ovarian cancer, familial 1, Hereditary cancer-predisposing syndrome
139877390	NM_001040431.2(COA3):c.215A>G (p.Tyr72Cys)	CCAYCTGGGGAGGTAGGTTCAGG	
793888527	NM_005859.4(PURA):c.563T>C (p.Ile188Thr)	GACCAYTGCGCTGCCCGCGCAGG, ACCAYTGCGCTGCCCGCGCAGGG, CCAYTGCGCTGCCCGCGCAGGGG	not provided, Mental retardation, autosomal dominant 31
561425038	NM_002878.3(RAD51D):c.1A>G (p.Met1Val)	CGCCCAYGTTCCCCGCAGGCCGG	Hereditary cancer-predisposing syndrome
121907934	NM_024105.3(ALG12):c.473T>C (p.Leu158Pro)	TCCYGCTGGCCCTCGCGGCCTGG	Congenital disorder of glycosylation type 1G
80358207	NM_153212.2(GJB4):c.409T>C (p.Phe137Leu)	CCTCATCYTCAAGGCCGCCGTGG	Erythrokeratodermia variabilis
80358228	NM_002353.2(TACSTD2):c.557T>C (p.Leu186Pro)	TCGGCYGCACCCCAAGTTCGTGG	Lattice corneal dystrophy Type III
121908076	NM_138691.2(TMC1):c.1543T>C (p.Cys515Arg)	AGGACCTYGCTGGGAAACAATGG, ACCTYGCTGGGAAACAATGGTGG, CCTYGCTGGGAAACAATGGTGGG	Deafness, autosomal recessive 7
121908089	NM_017838.3(NHP2):c.415T>C (p.Tyr139His)	GGAGGCTYACGATGAGTGCCTGG, GGCTYACGATGAGTGCCTGGAGG	Dyskeratosis congenita autosomal recessive 1, Dyskeratosis congenita, autosomal recessive 2
121908154	NM_001243133.1(NLRP3):c.926T>C (p.Phe309Ser)	GGTGCCTYTGACGAGCACATAGG	Familial cold urticaria, Chronic infantile neurological, cutaneous and articular syndrome
121908158	NM_001033855.2(DCLRE1C):c.2T>C (p.Met1Thr)	GGCGCTAYGAGTTCTTTCGAGGG, GCGCTAYGAGTTCTTTCGAGGGG	Histiocytic medullary reticulosis
796052870	NM_018129.3(PNPO):c.2T>C (p.Met1Thr)	CCCCCAYGACGTGCTGGCTGCGG, CCCCAYGACGTGCTGGCTGCGGG, CCCAYGACGTGCTGGCTGCGGGG	not provided
121908318	NM_020427.2(SLURP1):c.43T>C (p.Trp15Arg)	GCAGCCYGGAGCATGGGCTGTGG	Acroerythrokeratoderma
121908352	NM_022124.5(CDH23):c.5663T>C (p.Phe1888Ser)	CTCACCTYCAACATCACTGCGGG	Deafness, autosomal recessive 12
121908520	NM_000030.2(AGXT):c.613T>C (p.Ser205Pro)	CCTGTACYCGGGCTCCCAGAAGG	Primary hyperoxaluria, type I
121908618	NM_004273.4(CHST3):c.920T>C (p.Leu307Pro)	CGTGCYGGCCTCGCGCATGGTGG	Spondyloepiphyseal dysplasia with congenital joint dislocations
11694	NM_006432.3(NPC2):c.199T>C (p.Ser67Pro)	TATTCAGYCTAAAAGCAGCAAGG	Niemann-Pick disease type C2
121908739	NM_000022.2(ADA):c.320T>C (p.Leu107Pro)	CCTGCYGGCCAACTCCAAAGTGG	Severe combined immunodeficiency due to ADA deficiency
80359022	NM_000059.3(BRCA2):c.7958T>C (p.Leu2653Pro)	TGCYTCTTCAACTAAAATACAGG	Familial cancer of breast, Breast-ovarian cancer, familial 2
121908902	NM_003880.3(WISP3):c.232T>C (p.Cys78Arg)	AAAATCYGTGCCAAGCAACCAGG, AAATCYGTGCCAAGCAACCAGGG, AATCYGTGCCAAGCAACCAGGGG	Progressive pseudorheumatoid dysplasia
121908947	NM_006892.3(DNMT3B):c.808T>C (p.Ser270Pro)	CAAGTTCYCCGAGGTGAGTCCGG, AAGTTCYCCGAGGTGAGTCCGGG, AGTTCYCCGAGGTGAGTCCGGGG	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
121909028	NM_000492.3(CFTR):c.3857T>C (p.Phe1286Ser)	AGCCTYTGGAGTGATACCACAGG	Cystic fibrosis
121909135	NM_000085.4(CLCNKB):c.1294T>C (p.Tyr432His)	CTTTGTCYATGGTGAGTCTGGGG	Bartter syndrome type 3
121909143	NM_001300.5(KLF6):c.506T>C (p.Leu169Pro)	GGAGCYGCCCTCGCCAGGGAAGG	
121909182	NM_001089.2(ABCA3):c.302T>C (p.Leu101Pro)	GCACYTGTGATCAACATGCGAGG	Surfactant metabolism dysfunction, pulmonary, 3
121909200	NM_000503.5(EYA1):c.1459T>C (p.Ser487Pro)	CACTCYCGCTCATTCACTCCCGG	Melnick-Fraser syndrome

121909247	NM_004970.2(IGFALS):c.1618T>C (p.Cys540Arg)	GGACYGTGGCTGCCCTCTCAAGG	Acid-labile subunit deficiency
121909253	NM_005570.3(LMAN1):c.2T>C (p.Met1Thr)	AGAYGGCGGGATCCAGGCAAAGG	Combined deficiency of factor V and factor VIII, 1
121909385	NM_000339.2(SLC12A3):c.1868T>C (p.Leu623Pro)	CAACCYGGCCCTCAGCTACTCGG	Familial hypokalemia-hypomagnesemia
121909497	NM_002427.3(MMP13):c.224T>C (p.Phe75Ser)	TTCTYCGGCTTAGAGGTGACTGG	Spondyloepimetaphyseal dysplasia, Missouri type
121909508	NM_000751.2(CHRND):c.188T>C (p.Leu63Pro)	AACCYCATCTCCCTGGTGAGAGG	MYASTHENIC SYNDROME, CONGENITAL, 3B, FAST-CHANNEL
121909519	NM_001100.3(ACTA1):c.287T>C (p.Leu96Pro)	CGAGCYTCGCGTGGCTCCCGAGG	Nemaline myopathy 3
121909572	NM_000488.3(SERPINC1):c.667T>C (p.Ser223Pro)	TGGGTGYCCAATAAGACCGAAGG	Antithrombin III deficiency
121909677	NM_000821.6(GGCX):c.896T>C (p.Phe299Ser)	TATGTYCTCCTACGTCATGCTGG	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
121909727	NM_001018077.1(NR3C1):c.2209T>C (p.Phe737Leu)	CTATTGCYTCCAAACATTTTTGG	Glucocorticoid resistance, generalized
139573311	NM_000492.3(CFTR):c.1400T>C (p.Leu467Pro)	TTCACYTCTAATGGTGATTATGG, TCACYTCTAATGGTGATTATGGG	Cystic fibrosis
121912441	NM_000454.4(SOD1):c.341T>C (p.Ile114Thr)	CATCAYTGGCCGCACACTGGTGG	Amyotrophic lateral sclerosis type 1
121912446	NM_000454.4(SOD1):c.434T>C (p.Leu145Ser)	CGTTYGGCTTGTGGTGTAATTGG, GTTYGGCTTGTGGTGTAATTGGG	Amyotrophic lateral sclerosis type 1
121912463	NM_000213.3(ITGB4):c.1684T>C (p.Cys562Arg)	GGCCAGYGTGTGTGTGAGCCTGG	Epidermolysis bullosa with pyloric atresia
121912492	NM_002292.3(LAMB2):c.961T>C (p.Cys321Arg)	CCTCAACYGCGAGCAGTGTCAGG	Nephrotic syndrome, type 5, with or without ocular abnormalities
397516659	NM_001399.4(EDA):c.2T>C (p.Met1Thr)	GGCCAYGGGCTACCCGGAGGTGG	Hypohidrotic X-linked ectodermal dysplasia
111033589	NM_021044.2(DHH):c.485T>C (p.Leu162Pro)	GTTGCYGGCGCGCCTCGCAGTGG	46,XY gonadal dysgenesis, complete, dhh-related
111033622	NM_000206.2(IL2RG):c.343T>C (p.Cys115Arg)	TGGCYGTCAGTTGCAAAAAAAGG	X-linked severe combined immunodeficiency
121912613	NM_001041.3(SI):c.1859T>C (p.Leu620Pro)	ATGCYGGAGTTCAGTTTGTTTGG	Sucrase-isomaltase deficiency
121912619	NM_016180.4(SLC45A2):c.1082T>C (p.Leu361Pro)	GAGTTTCYCATCTACGAAAGAGG	Oculocutaneous albinism type 4
61750581	NM_000552.3(VWF):c.4837T>C (p.Ser1613Pro)	CTGCCYCTGATGAGATCAAGAGG	von Willebrand disease, type 2a
121912653	NM_000546.5(TP53):c.755T>C (p.Leu252Pro)	CATCCYCACCATCATCACACTGG	Li-Fraumeni syndrome 1
111033683	NM_000155.3(GALT):c.386T>C (p.Met129Thr)	AGGTCAYGTGCTTCCACCCCTGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033752	NM_000155.3(GALT):c.677T>C (p.Leu226Pro)	CAGGAGCYACTCAGGAAGGTGGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
121912729	NM_000039.1(APOA1):c.593T>C (p.Leu198Ser)	GCGCTYGGCCGCGCGCCTTGAGG	Familial visceral amyloidosis, Ostertag type
769452	NM_000041.3(APOE):c.137T>C (p.Leu46Pro)	AACYGGCACTGGGTCGCTTTTGG	
121912762	NM_016124.4(RHD):c.329T>C (p.Leu110Pro)	ACACYGTTCAGGTATTGGGATGG	
111033824	NM_000155.3(GALT):c.1138T>C (p.Ter380Arg)	CGCCYGACCACGCCGACCACAGG, GCCYGACCACGCCGACCACAGGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033832	NM_000155.3(GALT):c.980T>C (p.Leu327Pro)	TCCYGCGCTCTGCCACTGTCCGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
730881974	NM_000455.4(STK11):c.545T>C (p.Leu182Pro)	GGGAACCYGCTGCTCACCACCGG, AACCYGCTGCTCACCACCGGTGG	Hereditary cancer-predisposing syndrome
1064644	NM_000157.3(GBA):c.703T>C (p.Ser235Pro)	GGGYCACTCAAGGGACAGCCCGG	Gaucher disease
796052090	NM_138413.3(HOGA1):c.533T>C (p.Leu178Pro)	GGACCYGCCTGTGGATGCAGTGG	Primary hyperoxaluria, type III
121913141	NM_000208.2(INSR):c.779T>C (p.Leu260Pro)	CTACCYGGACGGCAGGTGTGTGG	Leprechaunism syndrome
121913272	NM_006218.2(PIK3CA):c.1258T>C (p.Cys420Arg)	GGAACACYGTCCATTGGCATGGG, GAACACYGTCCATTGGCATGGGG	Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi, Neoplasm of ovary, PIK3CA Related Overgrowth Spectrum
61751310	NM_000552.3(VWF):c.8317T>C (p.Cys2773Arg)	GCTCCYGCTGCTCTCCGACACGG	von Willebrand disease, type 2a
312262799	NM_024408.3(NOTCH2):c.1438T>C (p.Cys480Arg)	TTCACAYGTCTGTGCATGCCAGG	Alagille syndrome 2
121913570	NM_000426.3(LAMA2):c.7691T>C (p.Leu2564Pro)	ATCATTCYTTTGGGAAGTGGAGG, TCATTCYTTTGGGAAGTGGAGGG	Merosin deficient congenital muscular dystrophy
121913640	NM_000257.3(MYH7):c.1046T>C (p.Met349Thr)	AACTCCAYGTATAAGCTGACAGG	Familial hypertrophic cardiomyopathy 1, Cardiomyopathy
121913642	NM_000257.3(MYH7):c.1594T>C (p.Ser532Pro)	CATCATGYCCATCCTGGAAGAGG	Dilated cardiomyopathy 1S

119463996	NM_001079802.1(FKTN):c.527T>C (p.Phe176Ser)	GTAGTCTYTCATGAGAGGAGTGG	Limb-girdle muscular dystrophy-dystroglycanopathy, type C4
587776456	NM_002049.3(GATA1):c.1240T>C (p.Ter414Arg)	GCTCAYGAGGGCACAGAGCATGG	GATA-1-related thrombocytopenia with dyserythropoiesis
63750654	NM_000184.2(HBG2):c.-228T>C	ATGCAAAYATCTGTCTGAAACGG	Fetal hemoglobin quantitative trait locus 1
587776519	NM_001999.3(FBN2):c.3725-15A>G	AGCAYTGCAACCACATTGTCAGG	Congenital contractural arachnodactyly
78365220	NM_000402.4(G6PD):c.473T>C (p.Leu158Pro)	TGCCCYCCACCTGGGGTCACAGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
63750741	NM_000179.2(MSH6):c.1346T>C (p.Leu449Pro)	CTGGGGCYGGTATTCATGAAAGG	Hereditary Nonpolyposis Colorectal Neoplasms
587776914	NM_017565.3(FAM20A):c.590-2A>G	GTAATCYGCAAAGGAGGAGAAGG, TAATCYGCAAAGGAGGAGAAGGG	Enamel-renal syndrome
5030809	NM_000551.3(VHL):c.292T>C (p.Tyr98His)	CCCYACCCAACGCTGCCGCCTGG	Von Hippel-Lindau syndrome, Hereditary cancer-predisposing syndrome
199476132	m.5728T>C	CAATCYACTTCTCCCGCCGCCGG, AATCYACTTCTCCCGCCGCCGGG	Cytochrome-c oxidase deficiency, Mitochondrial complex I deficiency
62637012	NM_014336.4(AIPL1):c.715T>C (p.Cys239Arg)	CTGCCAGYGCCTGCTGAAGAAGG, CCAGYGCCTGCTGAAGAAGGAGG	Leber congenital amaurosis 4
199476199	NM_207352.3(CYP4V2):c.1021T>C (p.Ser341Pro)	AAACTGGYCCTTATACCTGTTGG, AACTGGYCCTTATACCTGTTGGG	Bietti crystalline corneoretinal dystrophy
587777183	NM_006702.4(PNPLA6):c.3053T>C (p.Phe1018Ser)	CCTYTAACCGCAGCATCCATCGG	Boucher Neuhauser syndrome
199476389	NM_000487.5(ARSA):c.899T>C (p.Leu300Ser)	GGTCTCTYGCGGTGTGGAAAGGG	Metachromatic leukodystrophy
199476398	NM_016599.4(MYOZ2):c.142T>C (p.Ser48Pro)	TTAYCCCATCTCAGTAACCGTGG	Familial hypertrophic cardiomyopathy 16
119456967	NM_001037633.1(SIL1):c.1370T>C (p.Leu457Pro)	TTGCYGAAGGAGCTGAGATGAGG	Marinesco-Sj\xc3\xb6gren syndrome
730882253	NM_006888.4(CALM1):c.268T>C (p.Phe90Leu)	GGCAYTCCGAGTCTTTGACAAGG	Long QT syndrome 14
587777283	NM_012338.3(TSPAN12):c.413A>G (p.Tyr138Cys)	TAATCCAYAATTTGTCATCCTGG	Exudative vitreoretinopathy 5
587777306	NM_015884.3(MBTPS2):c.1391T>C (p.Phe464Ser)	GCTYTGCTTTGGATGGACAATGG	Palmoplantar keratoderma, mutilating, with periorificial keratotic plaques, X-linked
56378716	NM_000250.1(MPO):c.752T>C (p.Met251Thr)	TCACTCAYGTTCATGCAATGGGG	Myeloperoxidase deficiency
587777390	NM_005026.3(PIK3CD):c.1246T>C (p.Cys416Arg)	GCAGGACYGCCCCATTGCCTGGG	Activated PI3K-delta syndrome
587777480	NM_003108.3(SOX11):c.178T>C (p.Ser60Pro)	TATGGYCCAAGATCGAACGCAGG	Mental retardation, autosomal dominant 27
587777663	NM_001288767.1(ARMC5):c.1379T>C (p.Leu460Pro)	GCCCGACYGCGGGATGCTGGTGG	Acth-independent macronodular adrenal hyperplasia 2
61753033	NM_000350.2(ABCA4):c.5819T>C (p.Leu1940Pro)	AAGGCYACATGAACTAACCAAGG	Stargardt disease, Stargardt disease 1, Cone-rod dystrophy 3
200488568	NM_002972.3(SBF1):c.4768A>G (p.Thr1590Ala)	CAGGCGYCCTCTTGCTCAGCCGG	Charcot-Marie-Tooth disease, type 4B3
132630274	NM_000377.2(WAS):c.809T>C (p.Leu270Pro)	CGGAGTCYGTTCTCCAGGGCAGG	Severe congenital neutropenia X-linked
132630308	NM_001399.4(EDA):c.181T>C (p.Tyr61His)	CTGCYACCTAGAGTTGCGCTCGG	Hypohidrotic X-linked ectodermal dysplasia
60934003	NM_170707.3(LMNA):c.1589T>C (p.Leu530Pro)	ACGGCTCYCATCAACTCCACTGG, CGGCTCYCATCAACTCCACTGGG, GGCTCYCATCAACTCCACTGGGG	Benign scapuloperoneal muscular dystrophy with cardiomyopathy
180177160	NM_000030.2(AGXT):c.1076T>C (p.Leu359Pro)	GGTGCYGCGGATCGGCCTGCTGG, GTGCYGCGGATCGGCCTGCTGGG	Primary hyperoxaluria, type I
180177222	NM_000030.2(AGXT):c.449T>C (p.Leu150Pro)	GTGCYGCTGTTCTTAACCCACGG, TGCYGCTGTTCTTAACCCACGGG	Primary hyperoxaluria, type I
180177254	NM_000030.2(AGXT):c.661T>C (p.Ser221Pro)	GCTCATCYCCTTCAGTGACAAGG	Primary hyperoxaluria, type I
180177264	NM_000030.2(AGXT):c.757T>C (p.Cys253Arg)	GGGGCYGTGACGACCAGCCCAGG	Primary hyperoxaluria, type I
180177293	NM_000030.2(AGXT):c.893T>C (p.Leu298Pro)	GTATCYGCATGGGCGCCTGCAGG	Primary hyperoxaluria, type I
376785840	NM_001282227.1(CECR1):c.1232A>G (p.Tyr411Cys)	GAAATCAYAGGACAAGCCTTTGG	Polyarteritis nodosa
587779393	NM_000257.3(MYH7):c.4937T>C (p.Leu1646Pro)	GAGCCYCCAGAGCTTGTTGAAGG	Myopathy, distal, 1
587779410	NM_012434.4(SLC17A5):c.500T>C (p.Leu167Pro)	ATTGTACYCAGAGCACTAGAAGG	Sialic acid storage disease, severe infantile type
587779513	NM_000090.3(COL3A1):c.2337+2T>C (p.Gly762_Lys779del)	AGGYAACCCTTAATACTACCTGG	Ehlers-Danlos syndrome, type 4
777539013	NM_020376.3(PNPLA2):c.757+2T>C	GAACGGYGCGCGGACCCGGGCGG, AACGGYGCGCGGACCCGGGCGGG	Neutral lipid storage disease with myopathy
34557412	NM_012452.2(TNFRSF13B):c.310T>C (p.Cys104Arg)	ACTTCYGTGAGAACAAGCTCAGG	Immunoglobulin A deficiency 2, Common variable immunodeficiency 2
796052970	NM_001165963.1(SCN1A):c.1094T>C (p.Phe365Ser)	CAAGCTYTGATACCTTCAGTTGG, AAGCTYTGATACCTTCAGTTGGG	not provided

724159989	NC_012920.1:m.7505T>C	CCTCCAYGACTTTTTCAAAAAGG	Deafness, nonsyndromic sensorineural, mitochondrial
796053222	NM_014191.3(SCN8A):c.4889T>C (p.Leu1630Pro)	CGTCYGATCAAAGGCGCCAAAGG, GTCYGATCAAAGGCGCCAAAGGG	not provided
118192127	NM_000540.2(RYR1):c.10817T>C (p.Leu3606Pro)	TACTACCYGGACCAGGTGGGTGG, ACTACCYGGACCAGGTGGGTGGG, CTACCYGGACCAGGTGGGTGGGG	Central core disease
118192170	NM_000540.2(RYR1):c.14693T>C (p.Ile4898Thr)	AGGCAYTGGGGACGAGATCGAGG	Malignant hyperthermia susceptibility type 1, Central core disease
121917703	NM_005247.2(FGF3):c.466T>C (p.Ser156Pro)	GTACGTGYCTGTGAACGGCAAGG, TACGTGYCTGTGAACGGCAAGGG	Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
690016549	NM_005211.3(CSF1R):c.2450T>C (p.Leu817Pro)	CCGCCYGCCTGTGAAGTGGATGG	Hereditary diffuse leukoencephalopathy with spheroids
690016552	NM_005211.3(CSF1R):c.2566T>C (p.Tyr856His)	GAATCCCYACCCTGGCATCCTGG	Hereditary diffuse leukoencephalopathy with spheroids
121917738	NM_001098668.2(SFTPA2):c.593T>C (p.Phe198Ser)	GGAGACTYCCGCTACTCAGATGG, GAGACTYCCGCTACTCAGATGGG	Idiopathic fibrosing alveolitis, chronic form
690016559	NM_005211.3(CSF1R):c.1957T>C (p.Cys653Arg)	AGCCYGTACCCATGGAGGTAAGG, GCCYGTACCCATGGAGGTAAGGG	Hereditary diffuse leukoencephalopathy with spheroids
690016560	NM_005211.3(CSF1R):c.2717T>C (p.Ile906Thr)	GCAGAYCTGCTCCTTCCTTCAGG	Hereditary diffuse leukoencephalopathy with spheroids
121917769	NM_003361.3(UMOD):c.376T>C (p.Cys126Arg)	GGCCACAYGTGTCAATGTGGTGG, GCCACAYGTGTCAATGTGGTGGG	Familial juvenile gout
121917773	NM_003361.3(UMOD):c.943T>C (p.Cys315Arg)	ATGGCACYGCCAGTGCAAACAGG	Glomerulocystic kidney disease with hyperuricemia and isosthenuria
121917818	NM_007255.2(B4GALT7):c.617T>C (p.Leu206Pro)	TGCYCTCCAAGCAGCACTACCGG	Ehlers-Danlos syndrome progeroid type
121917824	NM_021615.4(CHST6):c.827T>C (p.Leu276Pro)	GGACCYGGCGCGGGAGCCGCTGG	Macular corneal dystrophy Type I
121917848	NM_000452.2(SLC10A2):c.728T>C (p.Leu243Pro)	TTTCYTCTGGCTAGAATTGCTGG	Bile acid malabsorption, primary
121918006	NM_000478.4(ALPL):c.1306T>C (p.Tyr436His)	TGGACYATGGTGAGACCTCCAGG	Infantile hypophosphatasia
121918010	NM_000478.4(ALPL):c.979T>C (p.Phe327Leu)	CAAAGGCYTCTTCTTGCTGGTGG, GGCYTCTTCTTGCTGGTGGAAGG	Infantile hypophosphatasia
121918088	NM_000371.3(TTR):c.400T>C (p.Tyr134His)	CCCCYACTCCTATTCCACCACGG	
121918110	NM_001042465.1(PSAP):c.1055T>C (p.Leu352Pro)	GAAGCYGCCGAAGTCCCTGTCGG	Gaucher disease, atypical, due to saposin C deficiency
121918137	NM_003730.4(RNASET2):c.550T>C (p.Cys184Arg)	CCAGYGCCTTCCACCAAGCCAGG	Leukoencephalopathy, cystic, without megalencephaly
121918191	NM_001127628.1(FBP1):c.581T>C (p.Phe194Ser)	GGAGTYCATTTTGGTGGACAAGG	Fructose-biphosphatase deficiency
121918306	NM_006946.2(SPTBN2):c.758T>C (p.Leu253Pro)	ACCAAGCYGCTGGATCCCGAAGG, AAGCYGCTGGATCCCGAAGGTGG, AGCYGCTGGATCCCGAAGGTGGG	Spinocerebellar ataxia 5
121918505	NM_000141.4(FGFR2):c.799T>C (p.Ser267Pro)	AATGCCYCCACAGTGGTCGGAGG	Pfeiffer syndrome, Neoplasm of stomach
121918643	NM_003126.2(SPTA1):c.620T>C (p.Leu207Pro)	GTGGAGCYGGTAGCTAAAGAAGG, TGGAGCYGGTAGCTAAAGAAGGG	Hereditary pyropoikilocytosis, Elliptocytosis 2
121918646	NM_001024858.2(SPTB):c.604T>C (p.Trp202Arg)	CTCCAGCYGGAAGGATGGCTTGG	Spherocytosis type 2
121918648	NM_001024858.2(SPTB):c.6055T>C (p.Ser2019Pro)	ATGCCYCTGTGGCTGAGGCGTGG	
727504166	NM_000543.4(SMPD1):c.475T>C (p.Cys159Arg)	TGAGGCCYGTGGCCTGCTCCTGG, GAGGCCYGTGGCCTGCTCCTGGG	Niemann-Pick disease, type A, Niemann-Pick disease, type B
193922915	NM_000434.3(NEU1):c.1088T>C (p.Leu363Pro)	CAGCYATGGCCAGGCCCCAGTGG	Sialidosis, type II
727504419	NM_000501.3(ELN):c.889+2T>C	CAGGYAACATCTGTCCCAGCAGG, AGGYAACATCTGTCCCAGCAGGG	Supravalvar aortic stenosis
376395543	NM_000256.3(MYBPC3):c.26-2A>G	GAGACYGAAGGGCCAGGTGGAGG	Primary familial hypertrophic cardiomyopathy, Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
1169305	NM_000545.6(HNF1A):c.1720G>A (p.Gly574Ser)	GATGCYGGCAGGGTCCTGGCTGG, ATGCYGGCAGGGTCCTGGCTGGG, TGCYGGCAGGGTCCTGGCTGGGG	Maturity-onset diabetes of the young, type 3
730880130	NM_000527.4(LDLR):c.1468T>C (p.Trp490Arg)	CTACYGGACCGACTCTGTCCTGG, TACYGGACCGACTCTGTCCTGGG	Familial hypercholesterolemia
281860286	NM_018713.2(SLC30A10):c.500T>C (p.Phe167Ser)	GGCGCTTYCGGGGGGCCTCAGGG	Hypermanganesemia with dystonia, polycythemia and cirrhosis
730880306	NM_145693.2(LPIN1):c.1441+2T>C	AAGGYACCGCGGGCCTCGCGCGG, AGGYACCGCGGGCCTCGCGCGGG	Myoglobinuria, acute recurrent, autosomal recessive
74315452	NM_000454.4(SOD1):c.338T>C (p.Ile113Thr)	TTGCAYCATTGGCCGCACACTGG	Amyotrophic lateral sclerosis type 1
730880455	NM_000169.2(GLA):c.41T>C (p.Leu14Pro)	CGCGCYTGCGCTTCGCTTCCTGG	not provided
267606656	NM_054027.4(ANKH):c.1015T>C (p.Cys339Arg)	AGCTCYGTTTCGTGATGTTTTGG	Craniometaphyseal dysplasia, autosomal dominant

267606687	NM_033409.3(SLC52A3):c.1238T>C (p.Val413Ala)	AGTTACGYCAAGGTGATGCTGGG	Brown-Vialetto-Van laere syndrome
267606721	NM_001928.2(CFD):c.640T>C (p.Cys214Arg)	GGTGYGCGGGGGCGTGCTCGAGG, GTGYGCGGGGGCGTGCTCGAGGG	Complement factor d deficiency
267606747	NM_001849.3(COL6A2):c.2329T>C (p.Cys777Arg)	CGCCYGCGACAAGCCACAGCAGG	Ullrich congenital muscular dystrophy
431905515	NM_001044.4(SLC6A3):c.671T>C (p.Leu224Pro)	CTGCACCYCCACCAGAGCCATGG	Infantile Parkinsonism-dystonia
267606857	NM_000180.3(GUCY2D):c.2846T>C (p.Ile949Thr)	AGAGAYCGCCAACATGTCACTGG	Cone-rod dystrophy 6
267606880	NM_022489.3(INF2):c.125T>C (p.Leu42Pro)	GCTGCYCCAGATGCCCTCTGTGG	Focal segmental glomerulosclerosis 5
515726191	NM_015713.4(RRM2B):c.581A>G (p.Glu194Gly)	AACTCCTYCTACAGCAGCAAAGG	RRM2B-related mitochondrial disease
267606917	NM_004646.3(NPHS1):c.793T>C (p.Cys265Arg)	GCTGCCGYGCGTGGCCCGAGGGG, CTGCCGYGCGTGGCCCGAGGGGG	Finnish congenital nephrotic syndrome
267607104	NM_001199107.1(TBC1D24):c.751T>C (p.Phe251Leu)	CAAGTTCYTCCACAAGGTGAGGG, TTCYTCCACAAGGTGAGGGCCGG	Myoclonic epilepsy, familial infantile
267607182	NM_144631.5(ZNF513):c.1015T>C (p.Cys339Arg)	TGGGCGCYGCATGCGAGGAGAGG, CGCYGCATGCGAGGAGAGGCTGG	Retinitis pigmentosa 58
267607211	NM_000229.1(LCAT):c.508T>C (p.Trp170Arg)	TATGACYGGCGGCTGGAGCCCGG	Norum disease
267607215	NM_016269.4(LEF1):c.181T>C (p.Ser61Pro)	GAACGAGYCTGAAATCATCCCGG	Sebaceous tumors, somatic
587783580	NM_178151.2(DCX):c.683T>C (p.Leu228Pro)	AAAAAACYCTACACTCTGGATGG	Heterotopia
587783644	NM_004004.5(GJB2):c.107T>C (p.Leu36Pro)	GATCCYCGTTGTGGCTGCAAAGG	Hearing impairment
587783653	NM_005682.6(ADGRG1):c.1460T>C (p.Leu487Pro)	CCCTGCYCACCTGCCTTTCCTGG	Polymicrogyria, bilateral frontoparietal
587783863	NM_000252.2(MTM1):c.958T>C (p.Ser320Pro)	GGAAYCTTTAAAAAAAGTGAAGG	Severe X-linked myotubular myopathy
267607751	NM_000249.3(MLH1):c.453+2T>C	ATCACGGYAAGAATGGTACATGG, TCACGGYAAGAATGGTACATGGG	Hereditary Nonpolyposis Colorectal Neoplasms
119103227	NM_000411.6(HLCS):c.710T>C (p.Leu237Pro)	CTATCYTTCTCAGGGAGGGAAGG	Holocarboxylase synthetase deficiency
119103237	NM_005787.5(ALG3):c.211T>C (p.Trp71Arg)	GATTGACYGGAAGGCCTACATGG	Congenital disorder of glycosylation type 1D
398122806	NM_003172.3(SURF1):c.679T>C (p.Trp227Arg)	CCACYGGCATTATCGAGACCTGG	Congenital myasthenic syndrome, acetazolamide-responsive
80338747	NM_004525.2(LRP2):c.7564T>C (p.Tyr2522His)	GTACCTGYACTGGGCTGACTGGG	Donnai Barrow syndrome
398122838	NM_001271723.1(FBXO38):c.616T>C (p.Cys206Arg)	TTCCTYGTATCCCAATGCTAAGG	Distal hereditary motor neuronopathy 2D
398122989	NM_014495.3(ANGPTL3):c.883T>C (p.Phe295Leu)	ACAAAACYTCAATGAAACGTGGG	Hypobetalipoproteinemia, familial, 2
80338945	NM_004004.5(GJB2):c.269T>C (p.Leu90Pro)	GCTCCYAGTGGCCATGCACGTGG	Deafness, autosomal recessive 1A, Hearing impairment
80338956	NM_000334.4(SCN4A):c.2078T>C (p.Ile693Thr)	AAGATCAYTGGCAATTCAGTGGG, AGATCAYTGGCAATTCAGTGGGG, GATCAYTGGCAATTCAGTGGGGG	Hyperkalemic Periodic Paralysis Type 1, Paramyotonia congenita of von Eulenburg
267608131	NM_000179.2(MSH6):c.4001+2T>C	CGGYAACTAACTAACTATAATGG	Hereditary Nonpolyposis Colorectal Neoplasms
587784573	NM_004963.3(GUCY2C):c.2782T>C (p.Cys928Arg)	TCCCYGTGCTGCTGGAGTTGTGG, CCCYGTGCTGCTGGAGTTGTGGG	Meconium ileus
267608511	NM_003159.2(CDKL5):c.659T>C (p.Leu220Pro)	CCAACYTTTTACTATTCAGAAGG	Early infantile epileptic encephalopathy 2
373842615	NM_000118.3(ENG):c.1273-2A>G	CCGCCYGCGGGGATAAAGCCAGG, CGCCYGCGGGGATAAAGCCAGGG	Haemorrhagic telangiectasia 1
185492581	NM_000335.4(SCN5A):c.376A>G (p.Lys126Glu)	GAATCTYCACAGCCGCTCTCCGG	Brugada syndrome
200533370	NM_133499.2(SYN1):c.1699A>G (p.Thr567Ala)	GATGYCTGACGGGTAGCCTGTGG, ATGYCTGACGGGTAGCCTGTGGG	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, not specified
118203981	NM_148960.2(CLDN19):c.269T>C (p.Leu90Pro)	GCTCCYGGGCTTCGTGGCCATGG	Hypomagnesemia 5, renal, with ocular involvement
137853892	NM_001235.3(SERPINH1):c.233T>C (p.Leu78Pro)	GTCGCYAGGGCTCGTGTCGCTGG, TCGCYAGGGCTCGTGTCGCTGGG	Osteogenesis imperfecta type 10
118204024	NM_000263.3(NAGLU):c.142T>C (p.Phe48Leu)	GGCCGACYTCTCCGTGTCGGTGG	Mucopolysaccharidosis, MPS-III-B
690016563	NM_005211.3(CSF1R):c.1745T>C (p.Leu582Pro)	CAACCYGCAGTTTGGTGAGATGG	Hereditary diffuse leukoencephalopathy with spheroids
58380626	NM_000526.4(KRT14):c.1243T>C (p.Tyr415His)	CGCCACCYACCGCCGCCTGCTGG, CACCYACCGCCGCCTGCTGGAGG, ACCYACCGCCGCCTGCTGGAGGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
113994151	NM_207346.2(TSEN54):c.277T>C (p.Ser93Pro)	TTGAAGYCTCCCGCGGTGAGCGG, AAGYCTCCCGCGGTGAGCGGCGG	Pontocerebellar hypoplasia type 4
113994206	NM_004937.2(CTNS):c.473T>C (p.Leu158Pro)	TGGTCYGAGCTTCGACTTCGTGG	Cystinosis

62516109	NM_000277.1(PAH):c.638T>C (p.Leu213Pro)	CCACTTCYTGAAAAGTACTGTGG	Phenylketonuria
370011798	NM_001302946.1(TRNT1):c.668T>C (p.Ile223Thr)	GCAAYTGCAGAAAATGCAAAAGG	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
62517167	NM_000277.1(PAH):c.293T>C (p.Leu98Ser)	AAGATCTYGAGGCATGACATTGG	Mild non-PKU hyperphenylalanemia
12021720	NM_001918.3(DBT):c.1150G>A (p.Gly384Ser)	GACYCACAGAGCCCAATTTCTGG	Intermediate maple syrup urine disease type 2
104886289	NM_000495.4(COL4A5):c.4756T>C (p.Cys1586Arg)	TCCCCATYGTCCTCAGGGATGGG	Alport syndrome, X-linked recessive
370471013	NC_012920.1:m.5559A>G	CAACYTACTGAGGGCTTTGAAGG	Leigh disease
121434215	NM_000487.5(ARSA):c.410T>C (p.Leu137Pro)	GCCTTCCYGCCCCCCCATCAGGG	Metachromatic leukodystrophy, adult type
386134128	NM_000096.3(CP):c.1123T>C (p.Tyr375His)	ACACTACYACATTGCCGCTGAGG	Deficiency of ferroxidase
121434275	NM_001127328.2(ACADM):c.1136T>C (p.Ile379Thr)	GTGCAGAYACTTGGAGGCAATGG	Medium-chain acyl-coenzyme A dehydrogenase deficiency
121434276	NM_001127328.2(ACADM):c.742T>C (p.Cys248Arg)	CAGCGAYGTTCAGATACTAGAGG	Medium-chain acyl-coenzyme A dehydrogenase deficiency
121434284	NM_002225.3(IVD):c.134T>C (p.Leu45Pro)	ATGGGCYAAGCGAGGAGCAGAGG	ISOVALERIC ACIDEMIA, TYPE I
121434334	NM_005908.3(MANBA):c.1513T>C (p.Ser505Pro)	ATTACGYCCAGTCCTACAAATGG, TTACGYCCAGTCCTACAAATGGG, TACGYCCAGTCCTACAAATGGGG	Beta-D-mannosidosis
121434366	NM_000159.3(GCDH):c.883T>C (p.Tyr295His)	CGCCCGGYACGGCATCGCGTGGG, GCCCGGYACGGCATCGCGTGGGG	Glutaric aciduria, type 1
60715293	NM_000424.3(KRT5):c.541T>C (p.Ser181Pro)	GTTTGCCYCCTTCATCGACAAGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
121434409	NM_001003722.1(GLE1):c.2051T>C (p.Ile684Thr)	AAGGACAYTCCTGTCCCCAAGGG	Lethal arthrogryposis with anterior horn cell disease
121434434	NM_001287.5(CLCN7):c.2297T>C (p.Leu766Pro)	GGGCCYGCGGCACCTGGTGGTGG	Osteopetrosis autosomal recessive 4
121434455	NM_000466.2(PEX1):c.1991T>C (p.Leu664Pro)	GATGACCYTGACCTCATTGCTGG	Zellweger syndrome
199422317	NM_001099274.1(TINF2):c.862T>C (p.Phe288Leu)	CTGYTTCCCTTTAGGAATCTCGG	Aplastic anemia
104895221	NM_001065.3(TNFRSF1A):c.349T>C (p.Cys117Arg)	CTCTTCTYGCACAGTGGACCGGG	TNF receptor-associated periodic fever syndrome (TRAPS)
137854459	NM_000138.4(FBN1):c.4987T>C (p.Cys1663Arg)	GGGACAYGTTACAACACCGTTGG	Marfan syndrome
387907075	NM_024027.4(COLEC11):c.505T>C (p.Ser169Pro)	CAGCTGYCCTGCCAGGGCCGCGG, AGCTGYCCTGCCAGGGCCGCGGG, GCTGYCCTGCCAGGGCCGCGGGG, CTGYCCTGCCAGGGCCGCGGGGG	Carnevale syndrome
1048095	NM_000352.4(ABCC8):c.674T>C (p.Leu225Pro)	TGCYGTCCAAAGGCACCTACTGG	Permanent neonatal diabetes mellitus
796065347	NM_019074.3(DLL4):c.1168T>C (p.Cys390Arg)	GAAYGTCCCCCCAACTTCACCGG	Adams-Oliver syndrome, ADAMS-OLIVER SYNDROME 6
137852347	NM_000402.4(G6PD):c.1054T>C (p.Tyr352His)	AGGGYACCTGGACGACCCCACGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
74315327	NM_213653.3(HFE2):c.302T>C (p.Leu101Pro)	GGACCYCGCCTTCCATTCGGCGG	Hemochromatosis type 2A
137852579	NM_000044.3(AR):c.2033T>C (p.Leu678Pro)	GTCCYGGAAGCCATTGAGCCAGG	
137852636	NM_001166107.1(HMGCS2):c.520T>C (p.Phe174Leu)	CCCTCYTCAATGCTGCCAACTGG	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
137852661	NM_033163.3(FGF8):c.118T>C (p.Phe40Leu)	TTCCCTGYTCCGGGCTGGCCGGG	Kallmann syndrome 6
121912967	NM_005215.3(DCC):c.503T>C (p.Met168Thr)	AGCCCAYGCCAACAATCCACTGG	
137852806	NM_001039523.2(CHRNA1):c.901T>C (p.Phe301Leu)	TGTGYTCCTTCTGGTCATCGTGG	Myasthenic syndrome, congenital, fast-channel
137852850	NM_182760.3(SUMF1):c.463T>C (p.Ser155Pro)	GGCGACYCCTTTGTCTTTGAAGG	Multiple sulfatase deficiency
137852886	NM_000158.3(GBE1):c.671T>C (p.Leu224Pro)	AATGTACYACCAAGAATCAAAGG	Glycogen storage disease, type IV, GLYCOGEN STORAGE DISEASE IV, NONPROGRESSIVE HEPATIC
137852911	NM_000419.3(ITGA2B):c.641T>C (p.Leu214Pro)	CTGGTGCYTGGGGCTCCTGGCGG	Glanzmann thrombasthenia
137852948	NM_138694.3(PKHD1):c.10658T>C (p.Ile3553Thr)	GAGCCCAYTGAAATACGCTCAGG	Polycystic kidney disease, infantile type
137852964	NM_024960.4(PANK2):c.178T>C (p.Ser60Pro)	ATTGACYCAGTCGGATTCAATGG	
137853020	NM_006899.3(IDH3B):c.395T>C (p.Leu132Pro)	TGCGGCYGAGGTAGGTGGTCTGG, GCGGCYGAGGTAGGTGGTCTGGG	Retinitis pigmentosa 46
137853249	NM_033500.2(HK1):c.1550T>C (p.Leu517Ser)	GACTTCTYGGCCCTGGATCTTGG, TTCTYGGCCCTGGATCTTGGAGG	Hemolytic anemia due to hexokinase deficiency

137853270	NM_000444.5(PHEX):c.1664T>C (p.Leu555Pro)	AGCYCCAGAAGCCTTTCTTTTGG	Familial X-linked hypophosphatemic vitamin D refractory rickets
137853325	NM_003639.4(IKBKG):c.1249T>C (p.Cys417Arg)	TGGAGYGCATTGAGTAGGGCCGG	Hypohidrotic ectodermal dysplasia with immune deficiency, Hyper-IgM immunodeficiency, X-linked, with hypohidrotic ectodermal dysplasia
28932769	NM_002055.4(GFAP):c.1055T>C (p.Leu352Pro)	GGACCYGCTCAATGTCAAGCTGG	Alexander disease
397507439	NM_002769.4(PRSS1):c.116T>C (p.Val39Ala)	TACCAGGYGTCCCTGAATTCTGG	Hereditary pancreatitis
387906446	NM_000132.3(F8):c.1729T>C (p.Ser577Pro)	AAAGAAYCTGTAGATCAAAGAGG	Hereditary factor VIII deficiency disease
387906482	NM_000133.3(F9):c.1031T>C (p.Ile344Thr)	ACGAACAYCTTCCTCAAATTTGG	Hereditary factor IX deficiency disease
387906508	NM_000131.4(F7):c.983T>C (p.Phe328Ser)	GACGTYCTCTGAGAGGACGCTGG	Factor VII deficiency
387906532	NM_001040113.1(MYH11):c.3791T>C (p.Leu1264Pro)	GAAGCYGGAGGCGCAGGTGCAGG	Aortic aneurysm, familial thoracic 4
387906658	NM_002465.3(MYBPC1):c.2566T>C (p.Tyr856His)	CAAACCYATATCCGCAGAGTTGG	Distal arthrogryposis type 1B
387906701	NM_003491.3(NAA10):c.109T>C (p.Ser37Pro)	TGGCCTTYCCTGGCCCCAGGTGG, GGCCTTYCCTGGCCCCAGGTGGG	N-terminal acetyltransferase deficiency
387906717	NM_000377.2(WAS):c.881T>C (p.Ile294Thr)	GACTTCAYTGAGGACCAGGGTGG, ACTTCAYTGAGGACCAGGGTGGG	Severe congenital neutropenia X-linked
387906809	NM_000287.3(PEX6):c.1601T>C (p.Leu534Pro)	CTTCYGGGCCGGGACCGTGATGG, TTCYGGGCCGGGACCGTGATGGG	Peroxisome biogenesis disorder 4B
387906965	NM_024513.3(FYCO1):c.4127T>C (p.Leu1376Pro)	CAGCCYGATCCCCATCACTGTGG	Cataract, autosomal recessive congenital 2
387906967	NM_006147.3(IRF6):c.65T>C (p.Leu22Pro)	GCCYCTACCCTGGGCTCATCTGG	Van der Woude syndrome, Popliteal pterygium syndrome
387906982	NM_025132.3(WDR19):c.20T>C (p.Leu7Pro)	TCTCACYGCTAGAAAAGACTTGG	Asphyxiating thoracic dystrophy 5
387907072	NM_032446.2(MEGF10):c.2320T>C (p.Cys774Arg)	GGGCAGYGTACTTGCCGCACTGG	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant
137854499	NM_005502.3(ABCA1):c.6026T>C (p.Phe2009Ser)	GAGTYCTTTGCCCTTTTGAGAGG	Familial hypoalphalipoproteinemia
387907117	NM_000196.3(HSD11B2):c.1012T>C (p.Tyr338His)	CCGCCGCYATTACCCCGGCCAGG, CGCCGCYATTACCCCGGCCAGGG	Apparent mineralocorticoid excess
387907170	NM_004453.3(ETFDH):c.1130T>C (p.Leu377Pro)	CCAAAACYCACCTTTCCTGGTGG	
387907205	NM_033360.3(KRAS):c.211T>C (p.Tyr71His)	GGACCAGYACATGAGGACTGGGG, CCAGYACATGAGGACTGGGGAGG, CAGYACATGAGGACTGGGGAGGG	Cardiofaciocutaneous syndrome 2
387907240	NM_024110.4(CARD14):c.467T>C (p.Leu156Pro)	CAGCAGCYGCAGGAGCACCTGGG	Pityriasis rubra pilaris
387907282	NM_152296.4(ATP1A3):c.2431T>C (p.Ser811Pro)	TGCCATCYCACTGGCGTACGAGG	Alternating hemiplegia of childhood 2
387907361	NM_005120.2(MED12):c.3493T>C (p.Ser1165Pro)	AGGACYCTGAGCCAGGGGCCCGG	Ohdo syndrome, X-linked
28933970	NM_006194.3(PAX9):c.62T>C (p.Leu21Pro)	GGCCGCYGCCCAACGCCATCCGG	Tooth agenesis, selective, 3
137854472	NM_000138.4(FBN1):c.3128A>G (p.Lys1043Arg)	TGCACYTGCCGTGGGTGCAGAGG	
727504261	NM_000257.3(MYH7):c.2708A>G (p.Glu903Gly)	AGCGCYCCTCAGCATCTGCCAGG	Cardiomyopathy, not specified
81002853	NM_000059.3(BRCA2):c.476-2A>G	ACCACYGGGGGTAAAAAAAGGGG, TACCACYGGGGGTAAAAAAAGGG, ATACCACYGGGGGTAAAAAAAGG	Familial cancer of breast, Breast-ovarian cancer, familial 2, Hereditary cancer-predisposing syndrome
119473032	NM_021020.3(LZTS1):c.355A>G (p.Lys119Glu)	CCCTYCTCGGAGCCCTGTAGAGG	
193922801	NM_000540.2(RYR1):c.7043A>G (p.Glu2348Gly)	TTCYCCTCCACGCTCTCGCCTGG	not provided
36210419	NM_000218.2(KCNQ1):c.652A>G (p.Lys218Glu)	GCCCCTYGGAGCCCACGCAGAGG	Torsades de pointes, Cardiac arrhythmia
121964989	NM_000108.4(DLD):c.1483A>G (p.Arg495Gly)	TTCTCYAAAAGCTTCTGATAAGG	Maple syrup urine disease, type 3
28936669	NM_000095.2(COMP):c.1418A>G (p.Asp473Gly)	ATTGYCGTCGTCGTCGTCGCAGG	
28936696	NM_018488.2(TBX4):c.1592A>G (p.Gln531Arg)	GTACYGTAAGGAAGATTCTCGGG, GGTACYGTAAGGAAGATTCTCGG	Ischiopatellar dysplasia
121965077	NM_000137.2(FAH):c.1141A>G (p.Arg381Gly)	TCCYGGTCTGACCATTCCCCAGG	Tyrosinemia type I
794728203	NM_000138.4(FBN1):c.3344A>G (p.Asp1115Gly)	ACTCAYCAATATCTGCAAAATGG	Thoracic aortic aneurysms and aortic dissections
786205436	NM_003002.3(SDHD):c.275A>G (p.Asp92Gly)	GAATAGYCCATCGCAGAGCAAGG	Fatal infantile mitochondrial cardiomyopathy
72551317	NM_000784.3(CYP27A1):c.776A>G (p.Lys259Arg)	AGTCCACYTGGGGAGGAAGGTGG	Cholestanol storage disease

786205687	NM_016218.2(POLK):c.1385A>G (p.Asn462Ser)	ATTCACAYTCTTCAACTTAATGG	Malignant tumor of prostate
794728280	NM_000138.4(FBN1):c.7916A>G (p.Tyr2639Cys)	TGTTCAYACTGGAAGCCGGCGGG, CTGTTCAYACTGGAAGCCGGCGG	Thoracic aortic aneurysms and aortic dissections
28937317	NM_000335.4(SCN5A):c.3971A>G (p.Asn1324Ser)	GCAYTGACCACCACCTCAAGTGG	Long QT syndrome 3, Congenital long QT syndrome
786205854	NM_144499.2(GNAT1):c.386A>G (p.Asp129Gly)	CGGAGYCCTTCCACAGCCGCTGG	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1G
104893776	NM_000539.3(RHO):c.533A>G (p.Tyr178Cys)	GGATGYACCTGAGGACAGGCAGG	Retinitis pigmentosa 4
28937590	NM_001257342.1(BCS1L):c.232A>G (p.Ser78Gly)	GACACYGAGGTGCTGAGTACGGG, CGACACYGAGGTGCTGAGTACGG	GRACILE syndrome
104893866	NM_000320.2(QDPR):c.449A>G (p.Tyr150Cys)	TGCCGYACCCGATCATACCTGGG, ATGCCGYACCCGATCATACCTGG	Dihydropteridine reductase deficiency
587776590	NM_015629.3(PRPF31):c.527+3A>G	GACAYACCCCTGGGTGGTGGAGG, GCGGACAYACCCCTGGGTGGTGG	Retinitis pigmentosa 11
104894015	NM_000162.3(GCK):c.641A>G (p.Tyr214Cys)	GTAGYAGCAGGAGATCATCGTGG	Hyperinsulinemic hypoglycemia familial 3
202247823	NM_000532.4(PCCB):c.1606A>G (p.Asn536Asp)	ATATYTGCATGTTTTCTCCAAGG	Propionic acidemia
104894199	NM_000073.2(CD3G):c.1A>G (p.Met1Val)	CCAYGTCAGTCTCTGTCCTCCGG	Immunodeficiency 17
104894208	NM_001814.4(CTSC):c.857A>G (p.Gln286Arg)	CTCCYGAGGGCTTAGGATTGGGG, CCTCCYGAGGGCTTAGGATTGGG, ACCTCCYGAGGGCTTAGGATTGG	Papillon-Lef\xc3\xa8vre syndrome, Haim-Munk syndrome
104894211	NM_001814.4(CTSC):c.1040A>G (p.Tyr347Cys)	TCCTACAYAGTGGTACTCAGAGG	Papillon-Lef\xc3\xa8vre syndrome, Periodontitis, aggressive, 1
104894290	NM_000448.2(RAG1):c.2735A>G (p.Tyr912Cys)	CTGYACTGGCAGAGGGATTCTGG	Histiocytic medullary reticulosis
104894354	NM_000217.2(KCNA1):c.676A>G (p.Thr226Ala)	GCGYTTCCACGATGAAGAAGGGG, AGCGYTTCCACGATGAAGAAGGG, CAGCGYTTCCACGATGAAGAAGG	Episodic ataxia type 1
104894425	NM_014239.3(EIF2B2):c.638A>G (p.Glu213Gly)	AGTTGTCYCAATACCTGCTTTGG	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
104894450	NM_000270.3(PNP):c.383A>G (p.Asp128Gly)	ATAYCTCCAACCTCAAACTTGGG, GATAYCTCCAACCTCAAACTTGG	Purine-nucleoside phosphorylase deficiency
147394623	NM_024887.3(DHDDS):c.124A>G (p.Lys42Glu)	GGCACTYCTTGGCATAGCGACGG	Retinitis pigmentosa 59
60723330	NM_005557.3(KRT16):c.374A>G (p.Asn125Ser)	GCGGTCAYTGAGGTTCTGCATGG	Pachyonychia congenita, type 1, Palmoplantar keratoderma, nonepidermolytic, focal
104894634	NM_030665.3(RAI1):c.4685A>G (p.Gln1562Arg)	CTGCTGCYGTCGTCGTCGCTTGG	Smith-Magenis syndrome
104894730	NM_000363.4(TNNI3):c.532A>G (p.Lys178Glu)	CCTYCTTCACCTGCTTGAGGTGG, CCTCCTYCTTCACCTGCTTGAGG	Familial restrictive cardiomyopathy 1
104894816	NM_002049.3(GATA1):c.653A>G (p.Asp218Gly)	GTCCTGYCCCTCCGCCACAGTGG	GATA-1-related thrombocytopenia with dyserythropoiesis
794726773	NM_001165963.1(SCN1A):c.1662+3A>G	GTGCCAYACCTGGTGTGGGGAGG	Severe myoclonic epilepsy in infancy
104894861	NM_000202.6(IDS):c.404A>G (p.Lys135Arg)	AAAGACTYTTCCCACCGACATGG	Mucopolysaccharidosis, MPS-II
104894874	NM_000266.3(NDP):c.125A>G (p.His42Arg)	TGGYGCCTCATGCAGCGTCGAGG	
191205969	NM_002420.5(TRPM1):c.296T>C (p.Leu99Pro)	AAGCYCTTAATATCTGTGCATGG	Congenital stationary night blindness, type 1C
794727073	NM_019109.4(ALG1):c.1188-2A>G	TAAACYGCAGAGAGAACCAAGGG, GTAAACYGCAGAGAGAACCAAGG	Congenital disorder of glycosylation type 1K
281875236	NM_001004334.3(GPR179):c.659A>G (p.Tyr220Cys)	CCCACAYATCCATCTGCCTGCGG	Congenital stationary night blindness, type 1E
28939094	NM_015915.4(ATL1):c.1222A>G (p.Met408Val)	CACCCAYCTTCTTCACCCCTCGG	Spastic paraplegia 3
281875324	NM_005359.5(SMAD4):c.989A>G (p.Glu330Gly)	ATCCATTYCAAAGTAAGCAATGG	Juvenile polyposis syndrome, Hereditary cancer-predisposing syndrome
77173848	NM_000037.3(ANK1):c.-108T>C	GGGCCYGGCCCGCACGTCACAGG	Spherocytosis, type 1, autosomal recessive
150181226	NM_001159772.1(CANT1):c.671T>C (p.Leu224Pro)	CGTCYGTACGTGGGCGGCCTGGG, GCGTCYGTACGTGGGCGGCCTGG	Desbuquois syndrome
397514253	NM_000041.3(APOE):c.237-2A>G	CGCCCYGCGGCCGAGAGGGCGGG, GCGCCCYGCGGCCGAGAGGGCGG	Familial type 3 hyperlipoproteinemia
397514348	NM_000060.3(BTD):c.278A>G (p.Tyr93Cys)	GTTCAYAGATGTCAAGGTTCTGG	Biotinidase deficiency
397514415	NM_000060.3(BTD):c.1313A>G (p.Tyr438Cys)	GGCAYACAGCTCTTTGGATAAGG	Biotinidase deficiency
397514501	NM_007171.3(POMT1):c.430A>G (p.Asn144Asp)	GAGCATYCTCTGTTTCAAAGAGG	Limb-girdle muscular dystrophy-dystroglycanopathy, type C1
370382601	NM_174917.4(ACSF3):c.1A>G (p.Met1Val)	GGCAGCAYTGCACTGACAGGCGG	not provided
72554332	NM_000531.5(OTC):c.238A>G (p.Lys80Glu)	AAGGACTYCCCTTGCAATAAAGG	Ornithine carbamoyltransferase deficiency
397514599	NM_033109.4(PNPT1):c.1424A>G	GACTYCAGATGTAACTCTTATGG	Deafness, autosomal recessive 70

	(p.Glu475Gly)		
397514650	NM_000108.4(DLD):c.1444A>G (p.Arg482Gly)	GACTCYAGCTATATCTTCACAGG	Maple syrup urine disease, type 3
397514675	NM_003156.3(STIM1):c.251A>G (p.Asp84Gly)	TTCCACAYCCACATCACCATTGG	Myopathy with tubular aggregates
794728378	NM_000238.3(KCNH2):c.1913A>G (p.Lys638Arg)	ATCYTCTCTGAGTTGGTGTTGGG, GATCYTCTCTGAGTTGGTGTTGG	Cardiac arrhythmia
397514711	NM_002163.2(IRF8):c.238A>G (p.Thr80Ala)	AACCTCGYCTTCCAAGTGGCTGG	Autosomal dominant CD11C+/CD1C+ dendritic cell deficiency
397514729	NM_000388.3(CASR):c.85A>G (p.Lys29Glu)	CCCCCTYCTTTTGGGCTCGCTGG	Hypocalcemia, autosomal dominant 1, with bartter syndrome
397514743	NM_022114.3(PRDM16):c.2447A>G (p.Asn816Ser)	GCCGCCGYTTTGGCTGGCACGGG	Left ventricular noncompaction 8
397514757	NM_005689.2(ABCB6):c.508A>G (p.Ser170Gly)	TGGGCYGTTCCAAGACACCAGGG, GTGGGCYGTTCCAAGACACCAGG	Dyschromatosis universalis hereditaria 3
28940313	NM_152443.2(RDH12):c.677A>G (p.Tyr226Cys)	CACTGCGYAGGTGGTGACCCCGG	Leber congenital amaurosis 13
794728538	NM_000218.2(KCNQ1):c.1787A>G (p.Glu596Gly)	GTCTYCTACTCGGTTCAGGCGGG, TGTCTYCTACTCGGTTCAGGCGG	Cardiac arrhythmia
794728569	NM_000218.2(KCNQ1):c.605A>G (p.Asp202Gly)	AGGYCTGTGGAGTGCAGGAGAGG	Cardiac arrhythmia
794728573	NM_000218.2(KCNQ1):c.1515-2A>G	GCCYGCAGTGGAGAGAGGAGAGG	Cardiac arrhythmia
370874727	NM_003494.3(DYSF):c.3349-2A>G	CCGCCCYGGAGACACGAAGCTGG	Limb-girdle muscular dystrophy, type 2B
794728859	NM_198056.2(SCN5A):c.2788-2A>G	ACCYGTCGAGATAATGGGTCAGG	not provided
794728887	NM_198056.2(SCN5A):c.4462A>G (p.Thr1488Ala)	CCTCTGYCATGAAGATGTCCTGG	not provided
28940878	NM_000372.4(TYR):c.125A>G (p.Asp42Gly)	CTCCTGYCCCCGCTCCACGGTGG	Tyrosinase-negative oculocutaneous albinism
397515420	NM_172107.2(KCNQ2):c.1636A>G (p.Met546Val)	GCAYGACACTGCAGGGGGGTGGG, CGCAYGACACTGCAGGGGGGTGG, AACCGCAYGACACTGCAGGGGGG	Early infantile epileptic encephalopathy 7
397515428	NM_001410.2(MEGF8):c.7099A>G (p.Ser2367Gly)	GACYCCCGTGAAATGATTCCCGG	Carpenter syndrome 2
143601447	NM_201631.3(TGM5):c.122T>C (p.Leu41Pro)	TCAACCYCACCCTGTACTTCAGG	Peeling skin syndrome, acral type
397515519	NM_000207.2(INS):c.*59A>G	GGGCYTTATTCCATCTCTCTCGG	Permanent neonatal diabetes mellitus
397515523	NM_000370.3(TTPA):c.191A>G (p.Asp64Gly)	CAGGYCCAGATCGAAATCCCGGG, CCAGGYCCAGATCGAAATCCCGG	Ataxia with vitamin E deficiency
397515891	NM_000256.3(MYBPC3):c.1224-2A>G	TACTTGCYGTAGAACAGAAGGGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397516082	NM_000256.3(MYBPC3):c.927-2A>G	GTCCCYGTGTCCCGCAGTCTAGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397516138	NM_000257.3(MYH7):c.2206A>G (p.Ile736Val)	TATCAAYGAACTGTCCCTCAGGG, CTATCAAYGAACTGTCCCTCAGG	Familial hypertrophic cardiomyopathy 1, Cardiomyopathy, not specified
1154510	NM_002150.2(HPD):c.97G>A (p.Ala33Thr)	ATGACGYGGCCTGAATCACAGGG, AATGACGYGGCCTGAATCACAGG	4-Alpha-hydroxyphenylpyruvate hydroxylase deficiency
397516330	NM_000260.3(MYO7A):c.6439-2A>G	ATATCCYGGGGGAGCAGAAAGGG, GATATCCYGGGGGAGCAGAAAGG	Usher syndrome, type 1
72556271	NM_000531.5(OTC):c.482A>G (p.Asn161Ser)	CAGCCCAYTGATAATTGGGATGG	not provided
606231260	NM_023073.3(C5orf42):c.3290-2A>G	ATCYATCAAATACAAAAATTTGG	Orofaciodigital syndrome 6
587777521	NM_004817.3(TJP2):c.1992-2A>G	CAGCTCYGAGAAGAAACCACGGG, TCAGCTCYGAGAAGAAACCACGG	Progressive familial intrahepatic cholestasis 4
730880846	NM_000257.3(MYH7):c.617A>G (p.Lys206Arg)	CTTCYTGCTGCGGTCCCCAATGG	Cardiomyopathy
397517978	NM_206933.2(USH2A):c.12067-2A>G	TTCCCYGTAAGAAAATTAACAGG	Usher syndrome, type 2A, Retinitis pigmentosa 39
606231409	NM_000216.2(ANOS1):c.1A>G (p.Met1Val)	GCACCAYGGCTGCGGGTCGAGGG, GGCACCAYGGCTGCGGGTCGAGG	Kallmann syndrome 1
80356546	NM_003334.3(UBA1):c.1639A>G (p.Ser547Gly)	TGGCYTGTCACCCGGATATGTGG	Arthrogryposis multiplex congenita, distal, X-linked
80356584	NM_194248.2(OTOF):c.766-2A>G	GACCYGCAGGCAGGAGAAGGGGG, TGACCYGCAGGCAGGAGAAGGGG, CTGACCYGCAGGCAGGAGAAGGG, GCTGACCYGCAGGCAGGAGAAGG	Deafness, autosomal recessive 9
730880930	NM_000257.3(MYH7):c.1615A>G (p.Met539Val)	GGAACAYGCACTCCTCTTCCAGG	Cardiomyopathy
118203947	NM_013319.2(UBIAD1):c.355A>G (p.Arg119Gly)	TCCYGTCATCACTCTTTTTGTGG	Schnyder crystalline corneal dystrophy
60171927	NM_000526.4(KRT14):c.368A>G (p.Asn123Ser)	GCGGTCAYTGAGGTTCTGCATGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
199422248	NM_001363.4(DKC1):c.941A>G (p.Lys314Arg)	AATCYTGGCCCCATAGCAGATGG	Dyskeratosis congenita X-linked

72558467	NM_000531.5(OTC):c.929A>G (p.Glu310Gly)	TCCACTYCTTCTGGCTTTCTGGG, ATCCACTYCTTCTGGCTTTCTGG	not provided
72558478	NM_000531.5(OTC):c.988A>G (p.Arg330Gly)	ACTTTCYGTTTTCTGCCTCTGGG, CACTTTCYGTTTTCTGCCTCTGG	not provided
118204455	NM_000505.3(F12):c.158A>G (p.Tyr53Cys)	GGTGGYACTGGAAGGGGAAGTGG	
80357477	NM_007294.3(BRCA1):c.5453A>G (p.Asp1818Gly)	TTGYCCTCTGTCCAGGCATCTGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
121907908	NM_024426.4(WT1):c.1021A>G (p.Ser341Gly)	CGCYCTCGTACCCTGTGCTGTGG	Mesothelioma
121907926	NM_000280.4(PAX6):c.1171A>G (p.Thr391Ala)	GTGGYGCCCGAGGTGCCCATTGG	Optic nerve aplasia, bilateral
121908023	NM_024740.2(ALG9):c.860A>G (p.Tyr287Cys)	TTAYACAAAACAATGTTGAGTGG	Congenital disorder of glycosylation type 1L
121908148	NM_001243133.1(NLRP3):c.1880A>G (p.Glu627Gly)	ACAATYCCAGCTGGCTGGGCTGG	Familial cold urticaria
121908166	NM_006492.2(ALX3):c.608A>G (p.Asn203Ser)	CGGYTCTGGAACCAGACCTGGGG, GCGGYTCTGGAACCAGACCTGGG, TGCGGYTCTGGAACCAGACCTGG	Frontonasal dysplasia 1
121908184	NM_020451.2(SEPN1):c.1A>G (p.Met1Val)	CCCAYGGCTGCGGCTGGCGGCGG, CGGCCCAYGGCTGCGGCTGGCGG	Eichsfeld type congenital muscular dystrophy
121908258	NM_130468.3(CHST14):c.878A>G (p.Tyr293Cys)	AAGTCAYAGTGCACGGCACAAGG	Ehlers-Danlos syndrome, musculocontractural type
121908383	NM_001128425.1(MUTYH):c.1241A>G (p.Gln414Arg)	AAGCYGCTCTGAGGGCTCCCAGG	Neoplasm of stomach
121908580	NM_004328.4(BCS1L):c.148A>G (p.Thr50Ala)	GTGYGATCATGTAATGGCGCCGG	Mitochondrial complex III deficiency
121908584	NM_016417.2(GLRX5):c.294A>G (p.Gln98=)	CCTGACCYTGTCGGAGCTCCGGG	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive
121908635	NM_022817.2(PER2):c.1984A>G (p.Ser662Gly)	GCCACACYCTCTGCCTTGCCCGG	Advanced sleep phase syndrome, familial
121908655	NM_003839.3(TNFRSF11A):c.508A>G (p.Arg170Gly)	GGGTCYGCATTTGTCCGTGGAGG	Osteopetrosis autosomal recessive 7
29001653	NM_000539.3(RHO):c.886A>G (p.Lys296Glu)	CGCTCTYGGCAAAGAACGCTGGG, GCGCTCTYGGCAAAGAACGCTGG	Retinitis pigmentosa 4
56307355	NM_006502.2(POLH):c.1603A>G (p.Lys535Glu)	AGACTTTYCTGCTTAAAGAAGGG	Xeroderma pigmentosum, variant type
121908919	NM_002977.3(SCN9A):c.1964A>G (p.Lys655Arg)	CCTTTTCYTGTGTATTTGATTGG	Generalized epilepsy with febrile seizures plus, type 7, not specified
121908939	NM_006892.3(DNMT3B):c.2450A>G (p.Asp817Gly)	GACACGYCTGTGTAGTGCACAGG	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
121909088	NM_001005360.2(DNM2):c.1684A>G (p.Lys562Glu)	ACTYCTTCTCTTTCTCCTGAGGG, TACTYCTTCTCTTTCTCCTGAGG	Charcot-Marie-Tooth disease, dominant intermediate b, with neutropenia
120074112	NM_000483.4(APOC2):c.1A>G (p.Met1Val)	GCCCAYAGTGTCCAGAGACCTGG	Apolipoprotein C2 deficiency
121909239	NM_000314.6(PTEN):c.755A>G (p.Asp252Gly)	ATAYCACCACACACAGGTAACGG	Macrocephaly/autism syndrome
121909251	NM_198217.2(ING1):c.515A>G (p.Asn172Ser)	TGGYTGCACAGACAGTACGTGGG, CTGGYTGCACAGACAGTACGTGG	Squamous cell carcinoma of the head and neck
121909396	NM_001174089.1(SLC4A11):c.2518A>G (p.Met840Val)	GATCAYCTTCATGTAGGGCAGGG, AGATCAYCTTCATGTAGGGCAGG	Corneal dystrophy and perceptive deafness
121909533	NM_000034.3(ALDOA):c.386A>G (p.Asp129Gly)	CCAYCCAACCCTAAGAGAAGAGG	HNSHA due to aldolase A deficiency
128627255	NM_004006.2(DMD):c.835A>G (p.Thr279Ala)	TGACCGYGATCTGCAGAGAAGGG, CTGACCGYGATCTGCAGAGAAGG	Dilated cardiomyopathy 3B
116929575	NM_001085.4(SERPINA3):c.1240A>G (p.Met414Val)	GCTCAYGAAGAAGATGTTCTGGG, TGCTCAYGAAGAAGATGTTCTGG	
61748392	NM_004992.3(MECP2):c.410A>G (p.Glu137Gly)	CAACYCCACTTTAGAGCGAAAGG	Mental retardation, X-linked, syndromic 13
61748906	NM_001005741.2(GBA):c.667T>C (p.Trp223Arg)	CCCACTYGGCTCAAGACCAATGG	Gaucher disease, type 1
199473024	NM_000238.3(KCNH2):c.3118A>G (p.Ser1040Gly)	CTGCYCTCCACGTCGCCCCGGGG, CCTGCYCTCCACGTCGCCCCGGG, GCCTGCYCTCCACGTCGCCCCGG	Sudden infant death syndrome
794728365	NM_000238.3(KCNH2):c.1129-2A>G	GGACCYGCACCCGGGGAAGGCGG	Cardiac arrhythmia
72556293	NM_000531.5(OTC):c.548A>G (p.Tyr183Cys)	AGAGCTAYAGTGTTCCTAAAAGG	not provided
111033244	NM_000441.1(SLC26A4):c.1151A>G (p.Glu384Gly)	TGAATYCCTAAGGAAGAGACTGG	Pendred syndrome, Enlarged vestibular aqueduct syndrome
111033415	NM_000260.3(MYO7A):c.1344-2A>G	AGCYGCAGGGGCACAGGGATGGG, AAGCYGCAGGGGCACAGGGATGG	Usher syndrome, type 1
121912439	NM_000454.4(SOD1):c.302A>G (p.Glu101Gly)	AGAATCTYCAATAGACACATCGG	Amyotrophic lateral sclerosis type 1
111033567	NM_002769.4(PRSS1):c.68A>G (p.Lys23Arg)	ATCYTGTCATCATCATCAAAGGG, GATCYTGTCATCATCATCAAAGG	Hereditary pancreatitis
121912565	NM_000901.4(NR3C2):c.2327A>G (p.Gln776Arg)	TCATCYGTTTGCCTGCTAAGCGG	Pseudohypoaldosteronism type 1 autosomal dominant

121912574	NM_000901.4(NR3C2):c.2915A>G (p.Glu972Gly)	CCGACYCCACCTTGGGCAGCTGG	Pseudohypoaldosteronism type 1 autosomal dominant
121912589	NM_001173464.1(KIF21A):c.2839A>G (p.Met947Val)	ATTCAYATCTGCCTCCATGTTGG	Fibrosis of extraocular muscles, congenital, 1
111033661	NM_000155.3(GALT):c.253-2A>G	ATTCACCYACCGACAAGGATAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033669	NM_000155.3(GALT):c.290A>G (p.Asn97Ser)	GAAGTCGYTGTCAAACAGGAAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033682	NM_000155.3(GALT):c.379A>G (p.Lys127Glu)	TGACCTYACTGGGTGGTGACGGG, ATGACCTYACTGGGTGGTGACGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033786	NM_000155.3(GALT):c.950A>G (p.Gln317Arg)	CAGCYGCCAATGGTTCCAGTTGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
121912765	NM_001202.3(BMP4):c.278A>G (p.Glu93Gly)	CCTCCYCCCCAGACTGAAGCCGG	Microphthalmia syndromic 6
121912856	NM_000094.3(COL7A1):c.425A>G (p.Lys142Arg)	CACCYTGGGGACACCAGGTCGGG, TCACCYTGGGGACACCAGGTCGG	Epidermolysis bullosa dystrophica inversa, autosomal recessive
199474715	NM_152263.3(TPM3):c.505A>G (p.Lys169Glu)	CCAACTYACGAGCCACCTACAGG	Congenital myopathy with fiber type disproportion
199474718	NM_152263.3(TPM3):c.733A>G (p.Arg245Gly)	ATCYCTCAGCAAACTCAGCACGG	Congenital myopathy with fiber type disproportion
121912895	NM_001844.4(COL2A1):c.2974A>G (p.Arg992Gly)	CCTCYCTCACCACGTTGCCCAGG	Spondyloepimetaphyseal dysplasia Strudwick type
121913074	NM_000129.3(F13A1):c.851A>G (p.Tyr284Cys)	ATAGGCAYAGATATTGTCCCAGG	Factor xiii, a subunit, deficiency of
121913145	NM_000208.2(INSR):c.707A>G (p.His236Arg)	GCTGYGGCAACAGAGGCCTTCGG	Leprechaunism syndrome
312262745	NM_025137.3(SPG11):c.2608A>G (p.Ile870Val)	ACTTAYCCTGGGGAGAAGGATGG	Spastic paraplegia 11, autosomal recessive
121913682	NM_000222.2(KIT):c.2459A>G (p.Asp820Gly)	AGAAYCATTCTTGATGTCTCTGG	Mast cell disease, systemic
587776757	NM_000151.3(G6PC):c.230+4A>G	GTTCYTACCACTTAAAGACGAGG	Glycogen storage disease type 1A
61752063	NM_000330.3(RS1):c.286T>C (p.Trp96Arg)	TTCTTCGYGGACTGCAAACAAGG	Juvenile retinoschisis
367543065	NM_024549.5(TCTN1):c.221-2A>G	AGCAACYGCAGAAAAAAGAGGGG, CAGCAACYGCAGAAAAAAGAGGG	Joubert syndrome 13
5030773	NM_000894.2(LHB):c.221A>G (p.Gln74Arg)	CCACCYGAGGCAGGGGCGGCAGG	Isolated lutropin deficiency
199476092	NM_000264.3(PTCH1):c.2479A>G (p.Ser827Gly)	CGTTACYGAAACTCCTGTGTAGG	Gorlin syndrome, Holoprosencephaly 7, not specified
398123158	NM_000117.2(EMD):c.450-2A>G	CGTTCCCYGAGGCAAAAGAGGGG	not provided
199476103	RMRP:n.71A>G	ACTTYCCCCTAGGCGGAAAGGGG, GACTTYCCCCTAGGCGGAAAGGG, GGACTTYCCCCTAGGCGGAAAGG	Metaphyseal chondrodysplasia, McKusick type, Metaphyseal dysplasia without hypotrichosis
5030856	NM_000277.1(PAH):c.1169A>G (p.Glu390Gly)	CTCYCTGCCACGTAATACAGGGG, ACTCYCTGCCACGTAATACAGGG, AACTCYCTGCCACGTAATACAGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
5030860	NM_000277.1(PAH):c.1241A>G (p.Tyr414Cys)	GGGTCGYAGCGAACTGAGAAGGG, TGGGTCGYAGCGAACTGAGAAGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
587777055	NM_020988.2(GNAO1):c.521A>G (p.Asp174Gly)	GGATGYCCTGCTCGGTGGGCTGG	Early infantile epileptic encephalopathy 17
587777223	NM_024301.4(FKRP):c.1A>G (p.Met1Val)	CCGCAYGGGGCCGAAGTCTGGGG, GCCGCAYGGGGCCGAAGTCTGGG, AGCCGCAYGGGGCCGAAGTCTGG	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5
587777479	NM_003108.3(SOX11):c.347A>G (p.Tyr116Cys)	GTACTTGYAGTCGGGGTAGTCGG	Mental retardation, autosomal dominant 27
587777496	NM_020435.3(GJC2):c.-170A>G	TTGYTCCCCCCTCGGCCTCAGGG, ATTGYTCCCCCCTCGGCCTCAGG	Leukodystrophy, hypomyelinating, 2
587777507	NM_022552.4(DNMT3A):c.1943T>C (p.Leu648Pro)	CTCCYGGTGCTGAAGGACTTGGG, GCTCCYGGTGCTGAAGGACTTGG	Tatton-Brown-rahman syndrome
587777557	NM_018400.3(SCN3B):c.482T>C (p.Met161Thr)	AATCAYGATGTACATCCTTCTGG	Atrial fibrillation, familial, 16
587777569	NM_001030001.2(RPS29):c.149T>C (p.Ile50Thr)	GATAYCGGTTTCATTAAGGTAGG	Diamond-Blackfan anemia 13
587777657	NM_153334.6(SCARF2):c.190T>C (p.Cys64Arg)	CCACGYGCTGCGCTGGCTGGAGG	Marden Walker like syndrome
587777689	NM_005726.5(TSFM):c.57+4A>G	ACTTCYCACCGGGTAGCTCCCGG	Combined oxidative phosphorylation deficiency 3
796052005	NM_000255.3(MUT):c.329A>G (p.Tyr110Cys)	GCAYACTGGCGGATGGTCCAGGG, AGCAYACTGGCGGATGGTCCAGG	not provided
587777809	NM_144596.3(TTC8):c.115-2A>G	GTTCCYGGAAAGCATTAAGAAGG	Retinitis pigmentosa 51
587777878	NM_000166.5(GJB1):c.580A>G (p.Met194Val)	TAGCAYGAAGACGGTGAAGACGG	X-linked hereditary motor and sensory neuropathy
74315420	NM_001029871.3(RSPO4):c.194A>G (p.Gln65Arg)	CGTACYGGCGGATGCCTTCCCGG	Anonychia
180177219	NM_000030.2(AGXT):c.424-2A>G (p.Gly_142Gln145del)	AGGCCCYGAGGAAGCAGGGACGG	Primary hyperoxaluria, type I

367610201	NM_002693.2(POLG):c.1808T>C (p.Met603Thr)	CTCAYGGCACTTACCTGGGATGG	not provided
180177319	NM_012203.1(GRHPR):c.84-2A>G	TCACAGCYGCGGGGAAAGGGAGG	Primary hyperoxaluria, type II
796052068	NM_000030.2(AGXT):c.777-2A>G	GGTACCYGGAAGACACGAGGGGG, TGGTACCYGGAAGACACGAGGGG	Primary hyperoxaluria, type I
61754010	NM_000552.3(VWF):c.1583A>G (p.Asn528Ser)	TGCCAYTGTAATTCCCACACAGG	von Willebrand disease, type 2a
587778866	NM_000321.2(RB1):c.1927A>G (p.Lys643Glu)	ATTYCAATGGCTTCTGGGTCTGG	Retinoblastoma
74435397	NM_006331.7(EMG1):c.257A>G (p.Asp86Gly)	ATAYCTGGCCGCGCTTCCCCAGG	Bowen-Conradi syndrome
796052527	NM_000156.5(GAMT):c.1A>G (p.Met1Val)	CGCTCAYGCTGCAGGCTGGACGG	not provided
796052637	NM_172107.2(KCNQ2):c.848A>G (p.Lys283Arg)	GTACYTGTCCCCGTAGCCAATGG	not provided
724159963	NM_032228.5(FAR1):c.1094A>G (p.Asp365Gly)	GATAYCATACAGGAATGCTGGGG, AGATAYCATACAGGAATGCTGGG, TAGATAYCATACAGGAATGCTGG	Peroxisomal fatty acyl-coa reductase 1 disorder
587779722	NM_000090.3(COL3A1):c.1762-2A>G (p.Gly588_Gln605del)	CACCCYAAAGAAGAAGTGGTCGG	Ehlers-Danlos syndrome, type 4
118192102	m.8296A>G	TTTACAGYGGGCTCTAGAGGGGG	Diabetes-deafness syndrome maternally transmitted
727502787	NM_001077494.3(NFKB2):c.2594A>G (p.Asp865Gly)	CTGYCTTCCTTCACCTCTGCTGG	Common variable immunodeficiency 10
727503036	NM_000117.2(EMD):c.266-2A>G	AGCCYTGGGAAGGGGGGCAGCGG	Emery-Dreifuss muscular dystrophy 1, X-linked
690016544	NM_005861.3(STUB1):c.194A>G (p.Asn65Ser)	GGCCCGGYTGGTGTAATACACGG	Spinocerebellar ataxia, autosomal recessive 16
690016554	NM_005211.3(CSF1R):c.2655-2A>G	GTATCYGGGAGATAGGACAGAGG	Hereditary diffuse leukoencephalopathy with spheroids
118192185	NM_172107.2(KCNQ2):c.1A>G (p.Met1Val)	GCACCAYGGTGCCTGGCGGGAGG	Benign familial neonatal seizures 1
121917869	NM_012064.3(MIP):c.401A>G (p.Glu134Gly)	AGATCYCCACTGTGGTTGCCTGG	Cataract 15, multiple types
121918014	NM_000478.4(ALPL):c.1250A>G (p.Asn417Ser)	AGGCCCAYTGCCATACAGGATGG	Infantile hypophosphatasia
121918036	NM_000174.4(GP9):c.110A>G (p.Asp37Gly)	GCAGYCCACCCACAGCCCCATGG	Bernard-Soulier syndrome type C
121918089	NM_000371.3(TTR):c.379A>G (p.Ile127Val)	CGGCAAYGGTGTAGCGGCGGGGG, GCGGCAAYGGTGTAGCGGCGGGG	Amyloidogenic transthyretin amyloidosis
121918121	NM_000823.3(GHRHR):c.985A>G (p.Lys329Glu)	CGACTYGGAGAGACGCCTGCAGG	Isolated growth hormone deficiency type 1B
121918333	NM_015335.4(MED13L):c.6068A>G (p.Asp2023Gly)	ATATCAYCTAGAGGGAAGGGGGG, CATATCAYCTAGAGGGAAGGGGG	Transposition of great arteries
121918605	NM_001035.2(RYR2):c.12602A>G (p.Gln4201Arg)	CGCCAGCYGCATTTCAAAGATGG	Catecholaminergic polymorphic ventricular tachycardia
587781262	NM_002764.3(PRPS1):c.343A>G (p.Met115Val)	TAGCAYATTTGCAACAAGCTTGG	Charcot-Marie-Tooth disease, X-linked recessive, type 5, Deafness, high-frequency sensorineural, X-linked
121918608	NM_001161766.1(AHCY):c.344A>G (p.Tyr115Cys)	GCGGGYACTTGGTGTGGATGAGG	Hypermethioninemia with s-adenosylhomocysteine hydrolase deficiency
121918613	NM_000702.3(ATP1A2):c.1033A>G (p.Thr345Ala)	CTGYCAGGGTCAGGCACACCTGG	Familial hemiplegic migraine type 2
587781339	NM_000535.5(PMS2):c.904-2A>G	GCAGACCYGCACAAAATACAAGG	Hereditary cancer-predisposing syndrome
121918691	NM_001128177.1(THRB):c.1324A>G (p.Met442Val)	CTTCAYGTGCAGGAAGCGGCTGG	Thyroid hormone resistance, generalized, autosomal dominant
121918692	NM_001128177.1(THRB):c.1327A>G (p.Lys443Glu)	CCACCTYCATGTGCAGGAAGCGG	Thyroid hormone resistance, generalized, autosomal dominant
727504333	NM_000256.3(MYBPC3):c.2906-2A>G	CCGTTCYGTGGGTATAGAGTGGG, GCCGTTCYGTGGGTATAGAGTGG	Familial hypertrophic cardiomyopathy 4
786200910	NM_006204.3(PDE6C):c.1483-2A>G	CTTTCYGTTGAAATAAGGATGGG, TCTTTCYGTTGAAATAAGGATGG	Achromatopsia 5
281860296	NM_000551.3(VHL):c.586A>T (p.Lys196Ter)	GGTCTTYCTGCACATTTGGGTGG	Von Hippel-Lindau syndrome
730880444	NM_000169.2(GLA):c.370-2A>G	GTGAACCYGAAATGAGAGGGAGG	not provided
730880531	NM_000256.3(MYBPC3):c.1227-2A>G	GTACCYGGGTGGGGGCCGCAGGG, TGTACCYGGGTGGGGGCCGCAGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
267606643	NM_013411.4(AK2):c.494A>G (p.Asp165Gly)	TCAYCTTTCATGGGCTCTTTTGG	Reticular dysgenesis
267606705	NM_005188.3(CBL):c.1144A>G (p.Lys382Glu)	TATTTYACATAGTTGGAATGTGG	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
62642934	NM_000277.1(PAH):c.916A>G (p.Ile306Val)	GGCCAAYTTCCTGTAATTGGGGG, AGGCCAAYTTCCTGTAATTGGGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
267606782	NM_000117.2(EMD):c.1A>G (p.Met1Val)	TCCAYGGCGGGTGCGGGCTCAGG	Emery-Dreifuss muscular dystrophy, X-linked
267606820	NM_014053.3(FLVCR1):c.361A>G (p.Asn121Asp)	AGGCGTYGACCAGCGAGTACAGG	Posterior column ataxia with retinitis pigmentosa

730880805	NM_000257.3(MYH7):c.4664A>G (p.Glu1555Gly)	GCCCYCCTCGTGCTCCAGGGAGG, CTTGCCCYCCTCGTGCTCCAGGG	Cardiomyopathy
267606834	NM_138387.3(G6PC3):c.346A>G (p.Met116Val)	TGATCAYGCAGTGTCCAGAAGGG, GTGATCAYGCAGTGTCCAGAAGG	Dursun syndrome
267606851	NM_000175.3(GPI):c.1028A>G (p.Gln343Arg)	GTACYGGTCATAGGGCAGCATGG	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
515726182	NM_015713.4(RRM2B):c.190T>C (p.Trp64Arg)	TTCCTTCYGGACAGCAGAAGAGG	RRM2B-related mitochondrial disease
730881002	NM_002880.3(RAF1):c.1279A>G (p.Ser427Gly)	GCTGCYGCCCTCGCACCACTGGG, GGCTGCYGCCCTCGCACCACTGG	Rasopathy
267607030	NM_002977.3(SCN9A):c.29A>G (p.Gln10Arg)	AAGCTCYGAGGTCCTGGGGGAGG	Primary erythromelalgia
267607048	NM_007373.3(SHOC2):c.4A>G (p.Ser2Gly)	TACYCATGGTGACTCAAGCCTGG	Noonan-like syndrome with loose anagen hair, Rasopathy
587783486	NM_004380.2(CREBBP):c.3983-2A>G	GCAGCCCYAGGAAGTCCAGAAGG	Rubinstein-Taybi syndrome
730881357	NM_000051.3(ATM):c.3154-2A>G	AGCCYACGGGAAAAGAACTGTGG	Hereditary cancer-predisposing syndrome
398122404	NM_001256864.1(DNAJC6):c.801-2A>G	AGGTATCYGAAACAGAAGGTTGG	Parkinson disease 19, juvenile-onset
267607482	NM_001927.3(DES):c.1024A>G (p.Asn342Asp)	GAATCGTYCTGCAGGAGAGGGGG	Myofibrillar myopathy 1
796053439	NM_000391.3(TPP1):c.833A>G (p.Gln278Arg)	CAGGTACYGCACATCTAGACTGG	not provided
587783835	NM_000252.2(MTM1):c.550A>G (p.Arg184Gly)	GTTATTCYCCAATGGTGATTGGG	Severe X-linked myotubular myopathy
587783842	NM_000252.2(MTM1):c.629A>G (p.Asp210Gly)	TCATCAYCTGAGGCACGATACGG	Severe X-linked myotubular myopathy
267607777	NM_000249.3(MLH1):c.884+4A>G	TGCTACAYTACCTGAGGTACAGG	Hereditary Nonpolyposis Colorectal Neoplasms
33972047	NM_000518.4(HBB):c.59A>G (p.Asn20Ser)	CACGYTCACCTTGCCCCACAGGG, CCACGYTCACCTTGCCCCACAGG	alpha Thalassemia
730882004	NM_000546.5(TP53):c.709A>G (p.Met237Val)	ACACAYGTAGTTGTAGTGGATGG	Li-Fraumeni syndrome, Hereditary cancer-predisposing syndrome
730882052	NM_001231.4(CASQ1):c.731A>G (p.Asp244Gly)	GGCTTGYCTGGGATGGTCACAGG	Myopathy, vacuolar, with casq1 aggregates
80338959	NM_000334.4(SCN4A):c.4078A>G (p.Met1360Val)	GATCAYGATGGTGATGTCGAAGG	Hyperkalemic Periodic Paralysis Type 1
80338960	NM_000334.4(SCN4A):c.4108A>G (p.Met1370Val)	CCATCAYGGTGACCATGTTGAGG	Hyperkalemic Periodic Paralysis Type 1
80338962	NM_000334.4(SCN4A):c.4774A>G (p.Met1592Val)	TGTACAYGTTGACCACGATGAGG	Hyperkalemic Periodic Paralysis Type 1, Familial hyperkalemic periodic paralysis
398123062	NM_012160.4(FBXL4):c.1694A>G (p.Asp565Gly)	TATGYCCAGCTGCTGTAACCTGG	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
730882140	NM_001039550.1(DNAJB2):c.14A>G (p.Tyr5Cys)	GATCTCGYAGTAGGATGCCATGG	Charcot-Marie-Tooth disease, Charcot-Marie-Tooth disease, axonal, type 2T
796053522	NM_052859.3(RFT1):c.1222A>G (p.Met408Val)	GCAYCACAAAATTGTACCTGGGG, AGCAYCACAAAATTGTACCTGGG, CAGCAYCACAAAATTGTACCTGG	Congenital disorder of glycosylation type 1N
398123211	NM_000169.2(GLA):c.548-2A>G	AACCYGTATGAGAAAACAATGGG, TAACCYGTATGAGAAAACAATGG	Fabry disease
587784423	NM_006306.3(SMC1A):c.616-2A>G	AGCCYGTGCAAACAGGGGAATGG	Congenital muscular hypertrophy-cerebral syndrome
398123411	NM_000487.5(ARSA):c.1108-2A>G	GGCTCYGGGGGCAGAGTCAGGGG, GGGCTCYGGGGGCAGAGTCAGGG, AGGGCTCYGGGGGCAGAGTCAGG	Metachromatic leukodystrophy
398123429	NM_000512.4(GALNS):c.1171A>G (p.Met391Val)	CCGCCAYCAGCGTGTCGCCACGG	Mucopolysaccharidosis, MPS-IV-A
267608500	NM_003159.2(CDKL5):c.578A>G (p.Asp193Gly)	ATGYCCACGGACTTTCCATAGGG, CATGYCCACGGACTTTCCATAGG	Early infantile epileptic encephalopathy 2
398123552	NM_000402.4(G6PD):c.188T>C (p.Ile63Thr)	ACACACAYATTCATCATCATGGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
75391579	NM_000155.3(GALT):c.563A>G (p.Gln188Arg)	TTACCYGGCAGTGGGGGTGGGGG, CTTACCYGGCAGTGGGGGTGGGG, CCTTACCYGGCAGTGGGGGTGGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
398123639	NM_001848.2(COL6A1):c.805-2A>G	TTCTCCCYGGAACACAAAACAGG	Ullrich congenital muscular dystrophy, Bethlem myopathy
398123750	NM_003482.3(KMT2D):c.5645-2A>G	GCAGTTCYGTGGGGGAATGAAGG	Kabuki make-up syndrome
398124528	NM_144997.5(FLCN):c.1433-2A>G	CCCACYGGGGAGAAGGGCAGGGG, GCCCACYGGGGAGAAGGGCAGGG, GGCCCACYGGGGAGAAGGGCAGG	Hereditary cancer-predisposing syndrome
113994149	NM_025265.3(TSEN2):c.926A>G (p.Tyr309Cys)	CAGAGCAYAGACCAAGAAAAAGG	Pontocerebellar hypoplasia type 2B
281865052	NM_198578.3(LRRK2):c.5605A>G (p.Met1869Val)	TCAACAYAATATTTCTAGGCAGG	Parkinson disease 8, autosomal dominant
281865495	NM_004614.4(TK2):c.562A>G (p.Thr188Ala)	AAGYCTCAGGATTGGTCCGAAGG	Mitochondrial DNA depletion syndrome 2

756328339	NM_003494.3(DYSF):c.3041A>G (p.Tyr1014Cys)	CTAYACTCCCAGCCTGGGGGAGG, ATGCTAYACTCCCAGCCTGGGGG, GATGCTAYACTCCCAGCCTGGGG	Limb-girdle muscular dystrophy, type 2B
387906810	NM_153427.2(PITX2):c.262A>G (p.Lys88Glu)	TCTYGAACCAAACCTGGGGGCGG, GATTCTYGAACCAAACCTGGGGG, CGATTCTYGAACCAAACCTGGGG	Axenfeld-Rieger syndrome type 1
78310959	NM_030964.3(SPRY4):c.530A>G (p.Lys177Arg)	AGTGCYTGTCCAGCTCGGGTGGG, AAGTGCYTGTCCAGCTCGGGTGG	Hypogonadotropic hypogonadism 17 with or without anosmia
144109267	NM_207352.3(CYP4V2):c.1393A>G (p.Arg465Gly)	TTCCYGGGGCCAGCAGAGAAGGG, GTTCCYGGGGCCAGCAGAGAAGG	Bietti crystalline corneoretinal dystrophy
104886319	NM_000495.4(COL4A5):c.1340-2A>G	CACCYGAGTAAGATAAAGAAAGG	Alport syndrome, X-linked recessive
104886416	NM_000495.4(COL4A5):c.466-2A>G	ACCCYAAAAGAAGCCATCAATGG	Alport syndrome, X-linked recessive
121434443	NM_004984.2(KIF5A):c.827A>G (p.Tyr276Cys)	GAACAYAGCTTTTCTGGGGGAGG	Spastic paraplegia 10
199422314	NM_001099274.1(TINF2):c.850A>G (p.Thr284Ala)	TGACTGYGGGGCGCTCCTTATGG	Dyskeratosis congenita autosomal dominant
121434478	NM_004044.6(ATIC):c.1277A>G (p.Lys426Arg)	AGTGTACYTGACAGCAATGGTGG	AICAR transformylase/IMP cyclohydrolase deficiency
111033765	NM_000155.3(GALT):c.812A>G (p.Glu271Gly)	CGCYCAGCAGGGGTCAGCTCAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
121434606	NM_006006.4(ZBTB16):c.1849A>G (p.Met617Val)	GATCAYGGCCGAGTAGTCCCGGG, TGATCAYGGCCGAGTAGTCCCGG	Skeletal defects, genital hypoplasia, and mental retardation
566325901	NM_000017.3(ACADS):c.1108A>G (p.Met370Val)	AGCCCAYGCCGCCCAGGATCTGG	not provided
148665132	NM_012079.5(DGAT1):c.751+2T>C	ACCGCGGYGAGGACCTCTGTGGG	Diarrhea 7
111033830	NM_000155.3(GALT):c.574A>G (p.Ser192Gly)	TGCYGGCCCATACCTGTCAAGGG, CTGCYGGCCCATACCTGTCAAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
28933679	NM_000132.3(F8):c.5600A>G (p.His1867Arg)	GAGYGCACATCTTTTTCCTAGGG, TGAGYGCACATCTTTTTCCTAGG	Hereditary factor VIII deficiency disease
137852251	NM_000133.3(F9):c.917A>G (p.Asn306Ser)	GCTGCAYTGTAGTTGTGGTGAGG	Hereditary factor IX deficiency disease
141686175	NM_001287223.1(SCN11A):c.3473T>C (p.Leu1158Pro)	CGTGCGCYGTCCCAGTTTGAAGG	Episodic pain syndrome, familial, 3
137852331	NM_000402.4(G6PD):c.583A>G (p.Asn195Asp)	ATGCGGTYCCAGCCTCTGCTGGG	Favism, susceptibility to, Anemia, nonspherocytic hemolytic, due to G6PD deficiency
137852369	NM_000132.3(F8):c.5821A>G (p.Asn1941Asp)	TAGCCATYGATTGCTGGAGAAGG	Hereditary factor VIII deficiency disease
137852389	NM_000132.3(F8):c.398A>G (p.Tyr133Cys)	TCAYATTCAGCTCCTATAGCAGG	Hereditary factor VIII deficiency disease
137852406	NM_000132.3(F8):c.940A>G (p.Thr314Ala)	TGAGCAGYAAGGAAAGTTATTGG	Hereditary factor VIII deficiency disease
28931576	NM_000041.3(APOE):c.178A>G (p.Thr60Ala)	ACAGTGYCTGCACCCAGCGCAGG	
74315301	NM_000396.3(CTSK):c.990A>G (p.Ter330Trp)	GAGYCACATCTTGGGGAAGCTGG	Pyknodysostosis
137852540	NM_002764.3(PRPS1):c.341A>G (p.Asn114Ser)	TAGCATAYTTGCAACAAGCTTGG	Phosphoribosylpyrophosphate synthetase superactivity
137852624	NM_000215.3(JAK3):c.299A>G (p.Tyr100Cys)	AATCCTGYACAGCAGGACTTGGG	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-negative
137852640	NM_001166107.1(HMGCS2):c.500A>G (p.Tyr167Cys)	ACCACCGYAGCAGGCATTGGTGG	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
137852814	NM_005633.3(SOS1):c.1654A>G (p.Arg552Gly)	GCATCCYTTCCAGTGTACTCCGG	Noonan syndrome, Noonan syndrome 4, Rasopathy
137852865	NM_001171993.1(HPD):c.362A>G (p.Tyr121Cys)	CCTCAYATCCAGGCAAGAATTGG	4-Hydroxyphenylpyruvate dioxygenase deficiency
370898981	NM_138691.2(TMC1):c.1763+3A>G	TGGCCYACCAGATCATGCCTTGG	Deafness, autosomal recessive 7
118192167	NM_000540.2(RYR1):c.14387A>G (p.Tyr4796Cys)	CCATAYACCAGCCCAGGTACAGG	Malignant hyperthermia susceptibility type 1, Central core disease
137852972	NM_032667.6(BSCL2):c.263A>G (p.Asn88Ser)	CGAGACAYTGGCAACAGGGAAGG	Distal hereditary motor neuronopathy type 5, Silver spastic paraplegia syndrome, Charcot-Marie-Tooth disease, type 2
118192193	NM_172107.2(KCNQ2):c.356A>G (p.Glu119Gly)	CTTCYCATACTCCTTGATGGTGG, GCTCTTCYCATACTCCTTGATGG	Benign familial neonatal seizures 1
118192201	NM_172107.2(KCNQ2):c.622A>G (p.Met208Val)	GGATCAYCCGCAGAATCTGCAGG	Benign familial neonatal seizures 1
137853027	NM_001080463.1(DYNC2H1):c.9044A>G (p.Asp3015Gly)	ATAYCTCTAATTACATCAGGTGG, AGAATAYCTCTAATTACATCAGG	Short-rib thoracic dysplasia 3 with or without polydactyly
137853197	NM_144573.3(NEXN):c.1955A>G (p.Tyr652Cys)	ATAYACTCTCCTCCATCTTCTGG	Dilated cardiomyopathy 1CC, Cardiomyopathy, not specified
137853203	NM_000476.2(AK1):c.491A>G (p.Tyr164Cys)	TTCTCAYAGAAGGCGATGACGGG, TTTCTCAYAGAAGGCGATGACGG	Adenylate kinase deficiency, hemolytic anemia due to
786200859	NM_000308.2(CTSA):c.746+3A>G	TCCCAYACCTGTTCCCCAGAAGG	Galactosialidosis, adult

786200897	NM_003494.3(DYSF):c.1285-2A>G	CAGCYAGAAGACACAGGGAGGGG, ACAGCYAGAAGACACAGGGAGGG, CACAGCYAGAAGACACAGGGAGG	Limb-girdle muscular dystrophy, type 2B
786200928	NM_206933.2(USH2A):c.7595-2144A>G	CTCTTAYCTTGGGAAAGGAGAGG	Usher syndrome, type 2A
137853322	NM_003639.4(IKBKG):c.1219A>G (p.Met407Val)	CCAYATCAGGGGCCTGATACTGG	Incontinentia pigmenti syndrome
387906267	NM_000022.2(ADA):c.219-2A>G	CCCCYGGGAAGGGAAGAAAGGGG, GCCCCYGGGAAGGGAAGAAAGGG, AGCCCCYGGGAAGGGAAGAAAGG	Severe combined immunodeficiency due to ADA deficiency
387906362	NM_000492.3(CFTR):c.3717+4A>G	TCAAATCYCACCCTCTGGCCAGG	Cystic fibrosis
397507442	NM_002769.4(PRSS1):c.65A>G (p.Asp22Gly)	CTTGYCATCATCATCAAAGGGGG, TCTTGYCATCATCATCAAAGGGG, ATCTTGYCATCATCATCAAAGGG, GATCTTGYCATCATCATCAAAGG	Hereditary pancreatitis
137853971	NM_024598.3(USB1):c.502A>G (p.Arg168Gly)	CCACCYGGTTTTCTCTTGATTGG	Poikiloderma with neutropenia
2228063	NM_000067.2(CA2):c.754A>G (p.Asn252Asp)	TGTYCTTCAGTGGCTGAGCTGGG, CTGTYCTTCAGTGGCTGAGCTGG	
387906743	NM_001376.4(DYNC1H1):c.2909A>G (p.Tyr970Cys)	ATTCAAGYAGATTACCTGATTGG	Spinal muscular atrophy, lower extremity predominant 1, autosomal dominant
387906772	NM_002052.4(GATA4):c.928A>G (p.Met310Val)	TCCGCAYTGCAAGAGGCCTGGGG, TTCCGCAYTGCAAGAGGCCTGGG	Atrial septal defect 2
387906825	NM_000414.3(HSD17B4):c.650A>G (p.Tyr217Cys)	TGCCACAYACTCTGGCTTCAGGG	Gonadal dysgenesis with auditory dysfunction, autosomal recessive inheritance
387906895	NM_006587.3(CORIN):c.1414A>G (p.Ser472Gly)	GGATAACYTGTACTGTTGTAGGG	Preeclampsia/eclampsia 5
387906957	NM_016013.3(NDUFAF1):c.758A>G (p.Lys253Arg)	ACCYTGACCTCCTGCCAGTAGGG, TACCYTGACCTCCTGCCAGTAGG	Mitochondrial complex I deficiency
28933682	NM_000132.3(F8):c.5822A>G (p.Asn1941Ser)	TAGCCAYTGATTGCTGGAGAAGG	Hereditary factor VIII deficiency disease
387907135	NM_016464.4(TMEM138):c.389A>G (p.Tyr130Cys)	CAGYACAACACTGCTGCTGTGGG, GCAGYACAACACTGCTGCTGTGG	Joubert syndrome 16
137854530	NM_001077488.3(GNAS):c.1A>G (p.Met1Val)	GCCCAYGGCGGCGGCGGCGGCGG	Pseudohypoparathyroidism type 1A
387907176	NM_018105.2(THAP1):c.70A>G (p.Lys24Glu)	CCTCACTYGTGGAAAGAAACGGG	Dystonia 6, torsion
137854593	NM_000397.3(CYBB):c.1499A>G (p.Asp500Gly)	TCACAYCTTTCTCCTCATCATGG	Chronic granulomatous disease, X-linked
387907226	NM_000076.2(CDKN1C):c.832A>G (p.Lys278Glu)	CGCTYGGCGAAGAAATCTGCGGG, GCGCTYGGCGAAGAAATCTGCGG	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomalies
387907242	NM_022912.2(REEP1):c.304-2A>G	TCCYGTCAAAGGAAAAACAGAGG	Distal hereditary motor neuronopathy type 5B
387907291	NM_022787.3(NMNAT1):c.817A>G (p.Asn273Asp)	TGTYTCTCTGCAAAGGGGCCAGG	Leber congenital amaurosis 9
387907576	NM_001287.5(CLCN7):c.296A>G (p.Tyr99Cys)	TGTCAYAGTCCAAGCTCTGCAGG	Osteopetrosis autosomal dominant type 2, Osteopetrosis autosomal recessive 4

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Supplementary Figure 1-Uncropped gels with size/MW markers

Figure 1b:

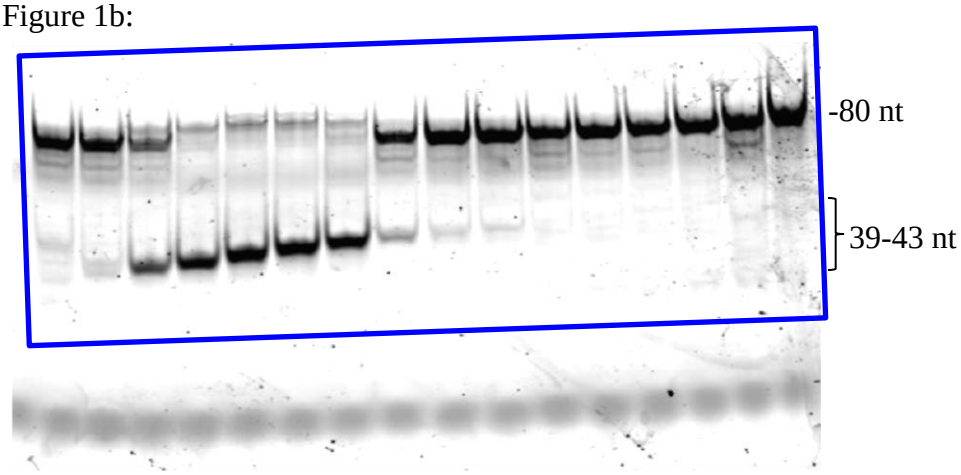
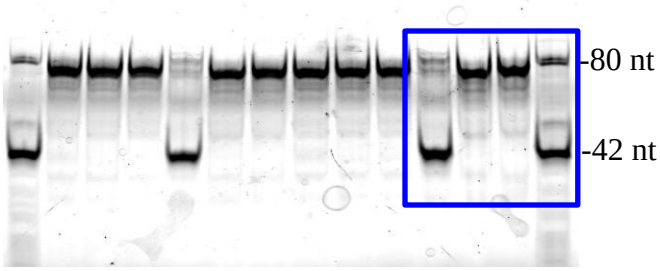
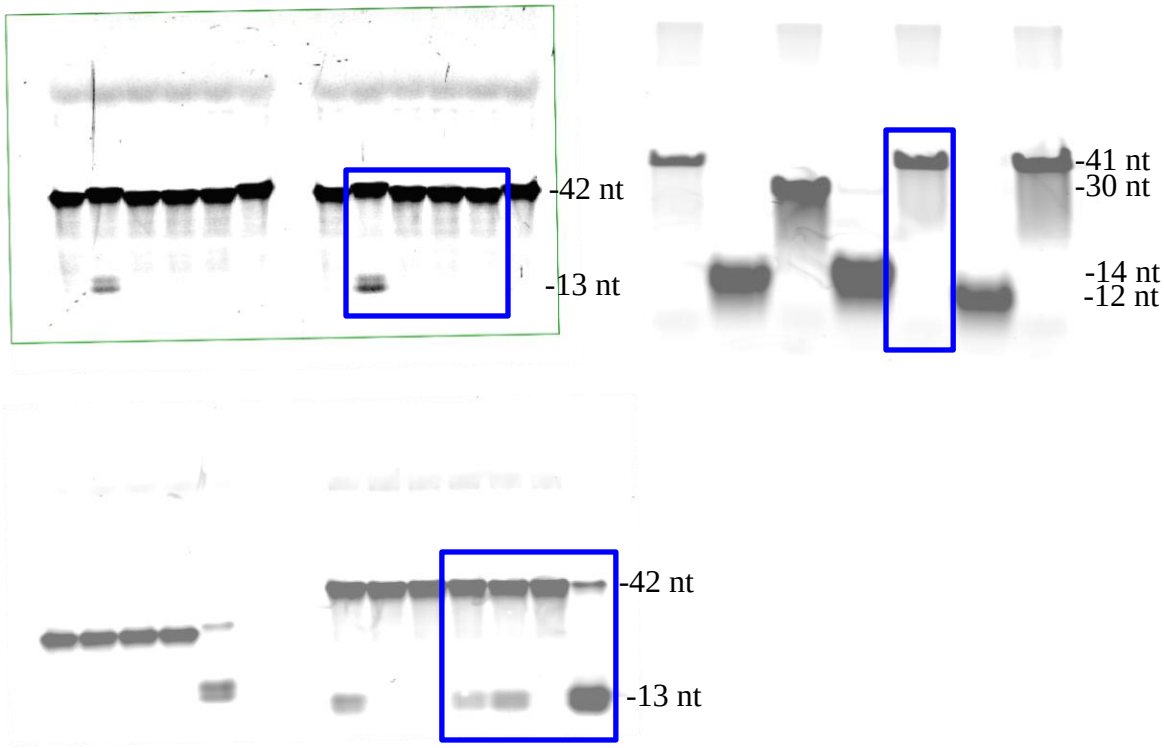


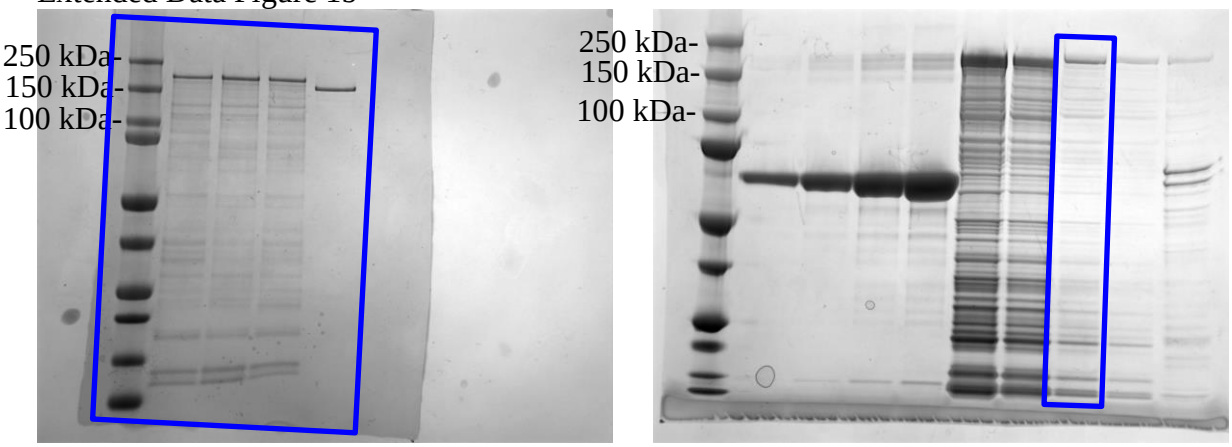
Figure 1c:



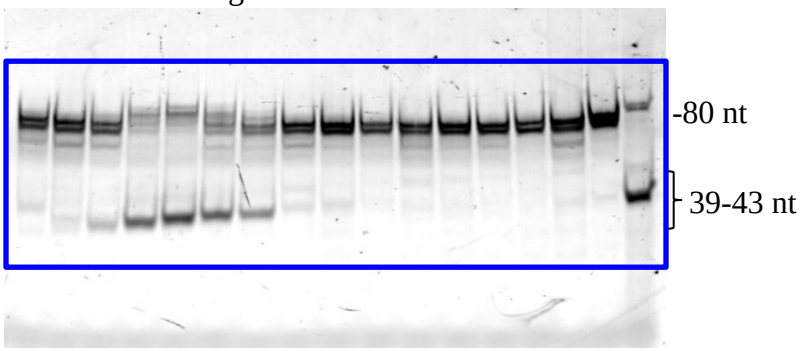
Extended Data Figure 1a:



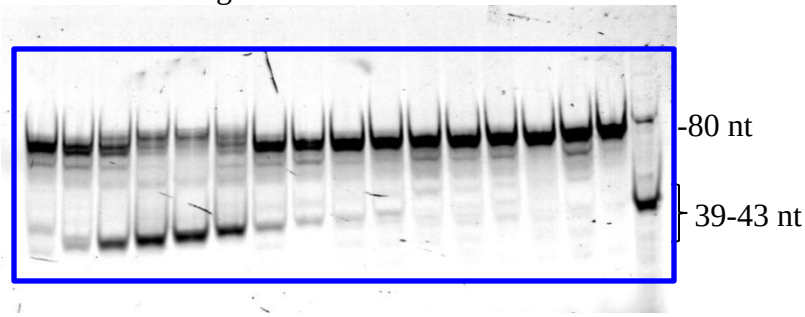
Extended Data Figure 1b



Extended Data Figure 1c:



Extended Data Figure 1d:



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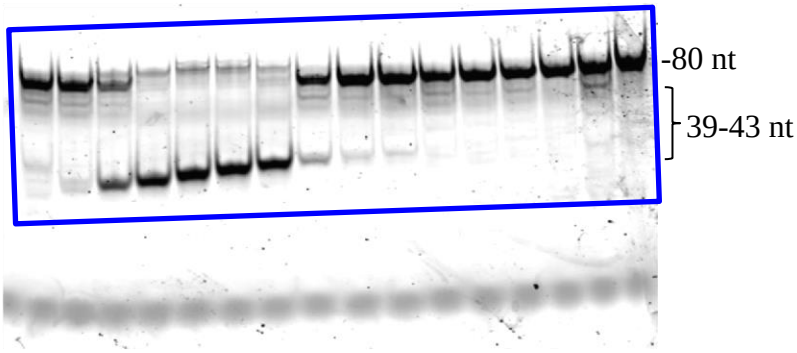

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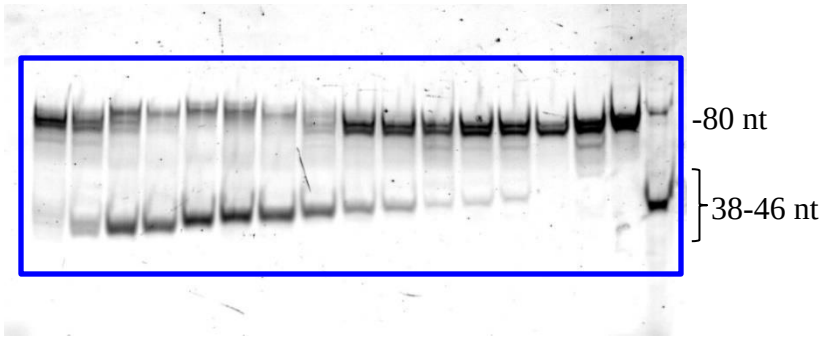

Extended Data Figure 2b: