Gene: GJB2 - Sequence: NG_008358.1
Transcript: NM_004004.5 - Protein: NP_003995.2
Date : March 2, 2015

1st line: Base numbering. Full stops for intronic +/- 5, 10, 15...
2nd line: Base sequence. lower case Introns, upper case Exons
3rd line: Amino acid sequence. Printed on FIRST base of codon
4th line: Amino acid numbering. Numbered on 1st and increments of 10

Exon 1 | Start: 5001 | End: 5193 | Length: 192

```
    .     .     .     .     .     .     .     .     .     .     .
ctggggggtcccgactctcagccgcccccgcttcacccgggccgccaaggggctggggga


    .     .     .     .     .     .     .     .     .     .     .
ggcggcgctcggggtaaccggggggagactcagggcgctgggggcacttggggaactcatg


    .     .     .     .     .     .     .     .     .     .     .
ggggctcaaaggaactaggagatcgggacctcgaagggggacttgggggggttcggggcttt


    .     .     .     .     .     .     .     .     .     .     .
cgggggcggtcggggggttcgcggacccgggaagctctgaggacccagaggccgggcgcgc


    .     .     .     .     .     .     .     .     .     .     .
tccgcccgcggcgccgccccctccgtaactttcccagtctccgagggaagaggcggggtg


        |-209      |-199      |-189      |-179      |-169      |-159
GGGGTGCGGTTAAAAGGCGCCACGGCGGGAGACAGGTGTTGCGGCCCCGCAGCGCCCGCG


        |-149      |-139      |-129      |-119      |-109      |-99
CGCTCCTCTCCCCGACTCGGAGCCCCTCGGCGGCGCCCGGCCCAGGACCCGCCTAGGAGC


        |-89       |-79       |-69       |-59       |-49       |-39
GCAGGAGCCCCAGCGCAGAGACCCCAACGCCGAGACCCCCGCCCCGGCCCCGCCGCGCTT


        |-29        .     .     .     .     .     .     .     .     .
CCTCCCGACGCAGgtgagcccgccggccccggactgcccggccaggaacctggcgcgggg


    .     .     .     .     .     .     .     .     .     .     .     .
agggaccgcgagacccagagcggttgcccggccgcgtgggtctcggggaaccgggggggct


    .     .     .     .     .     .     .     .     .     .     .     .
ggaccaacacacgtccttgggccgggggggcggggggccgccttctggagcgggcgtttctg
```

```
        .         .         .         .         .         .         .         .         .         .         .         .
cggccgagctccggagctggaatggggcggccggggaagtggacgcgatggcaccgcccg

        .         .         .         .         .         .         .         .         .         .         .         .
gggtgcgagtggggccgggcgcgcgcgggaggggaaaaaggcgcgggcgagccgccagcg

        .         .         .
cgaggtttgtggt
```

Exon 2 | Start: 8373 | End: 10513 | Length: 2140

```
   .      .      .      .      .      .      .      .      .      .      .      .
gacactccccagcacagcaaattttttatgatgtgtttaaagattgggtgaattactcagg

   .      .      .      .      .      .      .      .      .      .      .      .
tgaacaagctactttttatcagagaacacctaaaaacacgttcaagagggtttgggaact

   .      .      .      .      .      .      .      .      .      .      .      .
atacatttaatcctatgacaaactaagttggttctgtcttcacctgttttggtgaggttg

   .      .      .      .      .      .      .      .      .      .      .      .
tgtaagagttggtgtttgctcaggaagagatttaagcatgcttgcttacccagactcaga

   .      .      .      .      .      .      .      .      .      .      .      .
gaagtctccctgttctgtcctagctagtgattcctgtgttgtgtgcattcgtcttttcca

   |-19         |-9        |1           |11         |21         |31
AGCAAACCGCCCAGAGTAGAAGATGGATTGGGGCACGCTGCAGACGATCCTGGGGGGGTGT
                          M  D  W  G  T  L  Q  T  I  L  G  G  V
                          |1                              |11

   |41         |51        |61          |71         |81         |91
GAACAAACACTCCACCAGCATTGGAAAGATCTGGCTCACCGTCCTCTTCATTTTTCGCAT
 N  K  H  S  T  S  I  G  K  I  W  L  T  V  L  F  I  F  R  I
                          |21                             |31

   |101        |111       |121         |131        |141        |151
TATGATCCTCGTTGTGGCTGCAAAGGAGGTGTGGGGAGATGAGCAGGCCGACTTTGTCTG
 M  I  L  V  V  A  A  K  E  V  W  G  D  E  Q  A  D  F  V  C
                          |41                             |51

   |161        |171       |181         |191        |201        |211
CAACACCCTGCAGCCAGGCTGCAAGAACGTGTGCTACGATCACTACTTCCCCATCTCCCA
 N  T  L  Q  P  G  C  K  N  V  C  Y  D  H  Y  F  P  I  S  H
                          |61                             |71

   |221        |231       |241         |251        |261        |271
CATCCGGCTATGGGCCCTGCAGCTGATCTTCGTGTCCACGCCAGCGCTCCTAGTGGCCAT
 I  R  L  W  A  L  Q  L  I  F  V  S  T  P  A  L  L  V  A  M
                          |81                             |91
```

3

```
     |281        |291        |301        |311        |321        |331
GCACGTGGCCTACCGGAGACATGAGAAGAAGAGGAAGTTCATCAAGGGGGAGATAAAGAG
 H  V  A  Y  R  R  H  E  K  K  R  K  F  I  K  G  E  I  K  S
                       |101                               |111

     |341        |351        |361        |371        |381        |391
TGAATTTAAGGACATCGAGGAGATCAAAACCCAGAAGGTCCGCATCGAAGGCTCCCTGTG
 E  F  K  D  I  E  E  I  K  T  Q  K  V  R  I  E  G  S  L  W
                       |121                               |131

     |401        |411        |421        |431        |441        |451
GTGGACCTACACAAGCAGCATCTTCTTCCGGGTCATCTTCGAAGCCGCCTTCATGTACGT
 W  T  Y  T  S  S  I  F  F  R  V  I  F  E  A  A  F  M  Y  V
                       |141                               |151

     |461        |471        |481        |491        |501        |511
CTTCTATGTCATGTACGACGGCTTCTCCATGCAGCGGCTGGTGAAGTGCAACGCCTGGCC
 F  Y  V  M  Y  D  G  F  S  M  Q  R  L  V  K  C  N  A  W  P
                       |161                               |171

     |521        |531        |541        |551        |561        |571
TTGTCCCAACACTGTGGACTGCTTTGTGTCCCGGCCCACGGAGAAGACTGTCTTCACAGT
 C  P  N  T  V  D  C  F  V  S  R  P  T  E  K  T  V  F  T  V
                       |181                               |191

     |581        |591        |601        |611        |621        |631
GTTCATGATTGCAGTGTCTGGAATTTGCATCCTGCTGAATGTCACTGAATTGTGTTATTT
 F  M  I  A  V  S  G  I  C  I  L  L  N  V  T  E  L  C  Y  L
                       |201                               |211

     |641        |651        |661        |671        |681        |*11
GCTAATTAGATATTGTTCTGGGAAGTCAAAAAAGCCAGTTTAACGCATTGCCCAGTTGTT
 L  I  R  Y  C  S  G  K  S  K  K  P  V  *
                       |221

      |*21        |*31        |*41        |*51        |*61        |*71
AGATTAAGAAATAGACAGCATGAGAGGGATGAGGCAACCCGTGCTCAGCTGTCAAGGCTC

      |*81        |*91        |*101       |*111       |*121       |*131
AGTCGCTAGCATTTCCCAACACAAAGATTCTGACCTTAAATGCAACCATTTGAAACCCCT

      |*141       |*151       |*161       |*171       |*181       |*191
GTAGGCCTCAGGTGAAACTCCAGATGCCACAATGGAGCTCTGCTCCCCTAAAGCCTCAAA
```

4

```
   |*201        |*211        |*221        |*231        |*241        |*251
ACAAAGGCCTAATTCTATGCCTGTCTTAATTTTCTTTCACTTAAGTTAGTTCCACTGAGA

   |*261        |*271        |*281        |*291        |*301        |*311
CCCCAGGCTGTTAGGGGTTATTGGTGTAAGGTACTTTCATATTTTAAACAGAGGATATCG

   |*321        |*331        |*341        |*351        |*361        |*371
GCATTTGTTTCTTTCTCTGAGGACAAGAGAAAAAAGCCAGGTTCCACAGAGGACACAGAG

   |*381        |*391        |*401        |*411        |*421        |*431
AAGGTTTGGGTGTCCTCCTGGGGTTCTTTTTGCCAACTTTCCCCACGTTAAAGGTGAACA

   |*441        |*451        |*461        |*471        |*481        |*491
TTGGTTCTTTCATTTGCTTTGGAAGTTTTAATCTCTAACAGTGGACAAAGTTACCAGTGC

   |*501        |*511        |*521        |*531        |*541        |*551
CTTAAACTCTGTTACACTTTTTGGAAGTGAAAACTTTGTAGTATGATAGGTTATTTTGAT

   |*561        |*571        |*581        |*591        |*601        |*611
GTAAAGATGTTCTGGATACCATTATATGTTCCCCCTGTTTCAGAGGCTCAGATTGTAATA

   |*621        |*631        |*641        |*651        |*661        |*671
TGTAAATGGTATGTCATTCGCTACTATGATTTAATTTGAAATATGGTCTTTTGGTTATGA

   |*681        |*691        |*701        |*711        |*721        |*731
ATACTTTGCAGCACAGCTGAGAGGCTGTCTGTTGTATTCATTGTGGTCATAGCACCTAAC

   |*741        |*751        |*761        |*771        |*781        |*791
AACATTGTAGCCTCAATCGAGTGAGACAGACTAGAAGTTCCTAGTGATGGCTTATGATAG

   |*801        |*811        |*821        |*831        |*841        |*851
CAAATGGCCTCATGTCAAATATTTAGATGTAATTTTGTGTAAGAAATACAGACTGGATGT

   |*861        |*871        |*881        |*891        |*901        |*911
ACCACCAACTACTACCTGTAATGACAGGCCTGTCCAACACATCTCCCTTTTCCATGACTG

   |*921        |*931        |*941        |*951        |*961        |*971
TGGTAGCCAGCATCGGAAAGAACGCTGATTTAAAGAGGTCGCTTGGGAATTTTATTGACA

   |*981        |*991        |*1001       |*1011       |*1021       |*1031
CAGTACCATTTAATGGGGAGGACAAAATGGGGCAGGGGAGGGAGAAGTTTCTGTCGTTAA

   |*1041       |*1051       |*1061       |*1071       |*1081       |*1091
AAACAGATTTGGAAAGACTGGACTCTAAAGTCTGTTGATTAAAGATGAGCTTTGTCTACT
```

```
    |*1101     |*1111     |*1121     |*1131     |*1141     |*1151
TCAAAAGTTTGTTTGCTTACCCCTTCAGCCTCCAATTTTTTAAGTGAAAATATAGCTAAT

    |*1161     |*1171     |*1181     |*1191     |*1201     |*1211
AACATGTGAAAAGAATAGAAGCTAAGGTTTAGATAAATATTGAGCAGATCTATAGGAAGA

    |*1221     |*1231     |*1241     |*1251     |*1261     |*1271
TTGAACCTGAATATTGCCATTATGCTTGACATGGTTTCCAAAAAATGGTACTCCACATAT

    |*1281     |*1291     |*1301     |*1311     |*1321     |*1331
TTCAGTGAGGGTAAGTATTTTCCTGTTGTCAAGAATAGCATTGTAAAAGCATTTTGTAAT

    |*1341     |*1351     |*1361     |*1371     |*1381     |*1391
AATAAAGAATAGCTTTAATGATATGCTTGTAACTAAAATAATTTTGTAATGTATCAAATA

    |*1401     |*1411     |*1421     |*1431      .     .     .
CATTTAAAACATTAAAATATAATCTCTATAATAATTTAAAAtctaatatggtttttaatag

   .     .     .     .     .     .     .     .     .     .     .
aacagcaaattttaatttcatctatcactttttatataaatacattaatgtttttatattt

   .     .     .     .     .     .     .     .     .     .     .
cataacaccaatgggtaagttgccagagtgtctgaccccattctgccccagttacagaaa

   .     .     .     .     .     .     .     .     .     .     .
agcttctgtcaccagaaagtttggtggggaaggaagggaggaagatgatttctacctaac

   .     .     .     .     .     .     .     .     .     .     .
cccgtgcccacctctaccaggttttttgaggcatatcagtctatggacaatgtggtgtttg

   .     .     .     .     .     .     .     .     .
gtctggaaacgtaccttggtgaatgctgagttggctggaca
```

```
GBK Parser: Version: 1.1, Version Date: 11/02/2015
Reader: Version: 1, Version Date: 11/02/2015
Writer: Version: 1, Version Date: 11/02/2015
Control: Version: 1, Version Date: 11/02/2015
```