Supplementary Material

Supplementary Tables

Supplementary Table 1. The HNF1A SNPs associated with MODY3 or T2D.

| Location | Exon/Intron | Codon | Nucleotide and systematic name | Amino acid change | Diabetes type |
|----------|-------------|-------|---------------------------------|-------------------|---------------|
| Promoter | c.-538G>C   |       | {-538(G-C)}                     | MODY3             |
| Promoter | c.-373C>T   |       | {-373(C-T)}                     | MODY3             |
| Promoter | c.-283A>C   |       | {-283(A-C)}                     | MODY3             |
| Promoter | c.-218T>C   |       | {-218(T-C)}                     | MODY3             |
| Promoter | c.-158insGGTTGG |     | {-insGGTTGG}                  | MODY3             |
| Promoter | c.-58A>C    |       | {-58(A-C)}                      | MODY3             |
| Promoter | c.-58A>C    |       | {-58(A-C)}                      | MODY3             |
| Promoter | c.102G>C    |       | {102(G-C)}                      | MODY3             |
| Exon 1   | 6           | c.16A>C | p.S6R                           | MODY3             |
| Exon 1   | 7           | c.19C>T | p.Q7X                           | MODY3             |
| Exon 1   | 12          | c.35T>A | p.L12H                          | MODY3             |
| Exon 1   | 17          | c.51C>G | p.L17L                          | MODY3             |
| Exon 1   | 20          | c.59G>C | p.G20A                          | MODY3             |
| Exon 1   | 27          | c.79A>C | p.I27L                          | MODY3             |
| Exon 1   | 31          | c.91G>T | p.G31X                          | MODY3             |
| Exon 1   | 41          | c.121G>T | p.E41X                          | MODY3             |
| Exon 1   | 47          | c.140G>A | p.G47E                          | MODY3             |
| Exon 1   | 48          | c.142C>A | p.E48K                          | MODY3             |
| Exon 1   | 54          | c.160C>T | p.R54X                          | MODY3             |
| Exon 1   | 55/56       | c.161-165 delGAGGG | R55G56fsdelGAGG | MODY3             |
| Exon 1   | 58          | c.174T>G | p.A58A                          | MODY3             |
| Exon 1   | 74          | c.222G>T | p.T74T                          | MODY3             |
| Exon 1   | 80          | c.240C>G | p.D80E                          | MODY3             |
| Exon 1   | 98          | c.293C>T | p.A98V                          | MODY3             |
| Exon 1   | 103         | c.307G>A | p.V103M                         | MODY3             |
| Exon 1   | 107         | c.319C>G | p.L107I                         | MODY3             |
| Exon 2   | 112         | c.335C>T | p.P112L                         | MODY3             |
| Exon 2   | 114         | c.340C>T | p.R114C                         | MODY3             |
| Exon 2   | 122         | c.365A>G | p.Y122C                         | MODY3             |
| Exon 2   | 128         | c.383T>C | p.I128N                         | MODY3             |
| Exon 2   | 129         | c.385C>A | p.P129T                         | MODY3             |
| Exon 2   | 130         | c.388C>T | p.Q130X                         | MODY3             |
| Exon 2   | 131         | c.391C>T | p.R131W                         | MODY3             |
| Exon  | Position | Mutation | Amino Acid Change | Disease |
|-------|----------|----------|-------------------|---------|
| 2     | 131      | c.392G>A | p.R131Q          | MODY3   |
| 2     | 133      | c.397G>A | p.V133M          | MODY3   |
| 2     | 134      | c.402C>T | p.V134V          | MODY3   |
| 2     | 142      | c.425C>T | p.S142F          | MODY3   |
| 2     | 142      | c.426delG| S142fsdelG       | MODY3   |
| 2     | 143      | c.427C>T | p.H143Y          | MODY3   |
| 2     | 159      | c.476G>A | p.R159W          | MODY3   |
| 2     | 159      | c.476G>A | p.R159Q          | MODY3   |
| 2     | 162      | c.485T>C | p.L162P          | MODY3   |
| 2     | 171      | c.511C>T | p.R171X          | MODY3   |
| 2     | 171      | c.511C>T | p.R171X          | MODY3   |
| 2     | 171      | c.511C>T | p.R171X          | MODY3   |
| 2     | 196      | c.587_590delCCAA | T196fsdelCCAA | MODY3   |
| 3     | 203      | c.607C>G | p.R203G          | MODY3   |
| 3     | 229      | c.686G>A | p.R229Q          | MODY3   |
| 3     | 235      | c.703G>C | p.E235Q          | MODY3   |
| 3     | 241      | c.721T>G | p.C241G          | MODY3   |
| 3     | 245      | c.733G>C | p.G245R          | MODY3   |
| 3     | 260      | c.779C>T | p.T260M          | MODY3   |
| 3     | 263      | c.788G>A | p.R263H          | MODY3   |
| 3     | 269      | c.805G>C | p.A269P          | MODY3   |
| 4     | 271      | c.812G> | p.R271G          | MODY3   |
| 4     | 271      | c.812G>A | p.R271Q          | MODY3   |
| 4     | 271      | c.811C>T | p.R271W          | MODY3   |
| 4     | 272      | c.814C> | p.R272H          | MODY3   |
| 4     | 272      | c.814C>T | p.R272H          | MODY3   |
| 4     | 276      | c.827C>A | p.A276D          | MODY3   |
| 4     | 288      | c.864G>C | p.G288G          | MODY3   |
| 4     | 290      | c.869delC| Pro290fsdelC     | MODY3   |
| 4     | 291      | c.872delC| Pro291fsdelC     | MODY3   |
| 4     | 291      | c.872delA| Pro291fsdelA     | MODY3   |
| 4     | 291      | c.872delG| Pro291fsdelG     | MODY3   |
| 4     | 291      | c.873Insertion C | Pro291fsinsC | MODY3   |
| 4     | 291      | c.872dupC | Pro291dupC       | MODY3   |
| 4     | 291      | c.872^83insC | Pro291fsinsC | MODY3   |
| 4     | 292      | c.878delG | Pro292fsdelG     | MODY3   |
| 4     | 292      | c.901G>A | p.A301T          | MODY3   |
| 4     | 301      | c.956-2 A>G | c.956-2 A>G     | MODY3   |
| 4     | 321      | c.962 G>A | p.R321H          | MODY3   |
| 5     | 321      | c.962 G>A | p.R321H          | MODY3   |
| Exon | Position | Variant | Effect | Gene |
|------|----------|---------|--------|------|
| 5    | 349      | c.1047C>T | p.H349Q | MODY3 |
| 5    | 355      | c.1064C>A | p.S355X | MODY3 |
| 6    | 379      | c.1136^1137insC | P379fsinsC | MODY3 |
| 6    | 379      | c.1136-1137delT | P379fsdelT | MODY3 |
| 6    | 379      | c.1136-1137delCT | P379fsdelCT | MODY3 |
| 6    | 380      | c.1194T>C | p.Q398X | MODY3 |
| 6    | 414      | c.1238_1239dupCC | p.I414fs | MODY3 |
| 6    | 432      | c.1295C>G | p.S432Y | MODY3 |
| 6    | 433      | c.1328delCA | A443fsdelCA | MODY3 |
| 7    | 355      | c.1064C>A | p.S355X | MODY3 |
| 7    | 375      | c.1136^1137insC | P379fsinsC | MODY3 |
| 7    | 380      | c.1194T>C | p.Q398X | MODY3 |
| 7    | 414      | c.1238_1239dupCC | p.I414fs | MODY3 |
| 7    | 432      | c.1295C>G | p.S432Y | MODY3 |
| 7    | 433      | c.1328delCA | A443fsdelCA | MODY3 |
| 7    | 443      | c.1328delCA | A443fsdelCA | MODY3 |
| 7    | 451      | c.1340C>G | p.S432Y | MODY3 |
| 7    | 452      | c.1340C>G | p.S432Y | MODY3 |
| 7    | 459      | c.1375C>T | p.P447L | MODY3 |
| 7    | 466      | c.1396G>A | p.Q446* | MODY3 |
| 7    | 473      | c.1417C>T | p.Q473X | MODY3 |
| 7    | 487      | c.1460A>G | p.S487N | MODY3 |
| 7    | 488      | c.1463delC | P488fsdelC | MODY3 |
| 7    | 495      | c.1483C>T | p.Q495X | MODY3 |
| IVS7 |          | c.1501+7A>G | c.1501+7A>G | MODY3 |
| 8    | 519      | c.1556C>T | p.P519L | MODY3 |
| 8    | 521      | c.1562C>T | p.T521I | MODY3 |
| 8    | 531      | c.1592C>T | p.S531T | MODY3 |
| 8    | 547/548  | c.1641-1642delTG | T547E548fsdelTG | MODY3 |
| 9    | 559      | c.1677^1678insA | A559fsinsA | MODY3 |
| 9    | 584      | c.1752^1753insTC | L584S585fsinsTC | MODY3 |
| 9    | 584      | c.1764-1765insGCCA | P588fs-insGCCA | MODY3 |
| 9    | 588      | c.1764delAC | P588fs-delAC | MODY3 |
| Intron 9 | Splice site | c.1768+1G>A | c.1768+1G>A | MODY3 |
| 10   | 617      | c.1849G>A | p.V617I | MODY3 |
| 10   | 618      | c.1854C>G | p.I618M | MODY3 |
| 10   | 620      | c.1859C>T | p.T620I | MODY3 |
| 1    | 31       | c.92G>A | p.G31D | T2D |
| 1    | 48       | c.142G>A | p.E48K | T2D |
| 1    | 62       | c.185A>G | p.N62S | T2D |
| 1    | 97       | c.290C>T | p.A97V | T2D |
| 1    | 100      | c.298C>A | p.Q100K | T2D |
| 2    | 114      | c.341G>A | p.R114H | T2D |
| 2    | 131      | c.392G>A | p.R131Q | T2D |
| 2    | 141      | c.422_423insT | p.Q141Hfs *47 | T2D |
| 3    | 180      | c.539C>T | p.Ala180Val | T2D |
| 3    | 191      | c.572G>A | p.G191D | T2D |
| 3    | 196      | c.586A>G | p.T196A | T2D |
| 4    | 254      | c.760C>A | p.L254M | T2D |
| 4    | 263      | c.787C>T | p.R263C | T2D |
| Exon  | Base | Change | Protein Change | Condition |
|-------|------|--------|----------------|-----------|
| Exon 4 | 275  | c.825del | p.E275del | T2D |
| Exon 4 | 319  | c.955G>A*  | p.G319S | T2D |
| Exon 5 | 322  | c.965A>G  | p.Y322C | T2D |
| Exon 6 | 389  | c.1165T>G  | p.L389V | T2D |
| Exon 7 | 469  | c.1405C>T  | p.H469Y | T2D |
| Exon 7 | 487  | c.1460G>A  | p.S487N | T2D |
| Exon 8 | 505  | c.1513C>A  | p.H505N | T2D |
| Exon 8 | 508  | c.1522G>A# | p.E508K | T2D |
| Exon 8 | 514  | c.1541A>G  | p.H514R | T2D |
| Exon 8 | 515  | c.1544C>A  | p.T515K | T2D |
| Exon 8 | 537  | c.1610C>G  | p.T537R | T2D |
| Exon 9 | 566  | c.1696C>A  | p.H566N | T2D |
| Exon 9 | 574  | c.1720G>A  | p.G574S | T2D |
| Exon 9 | 577  | c.1729C>G  | p.H577D | T2D |
| Exon 9 | 583  | c.1748G>A  | p.R583Q | T2D |
| Exon 10| 619  | c.1856G>A  | p.E619K | T2D |