Supplementary Material

# Search strategy for database as follows.

# #1 "maturity onset diabetes of the young, type 3"[Supplementary Concept]

# #2 (((((Maturity-Onset Diabetes of the Young, Type 3[Title/Abstract]) OR (MODY 3[Title/Abstract])) OR (MODY3[Title/Abstract])) OR (HNF1A MODY[Title/Abstract])) OR (HNF1-alpha MODY[Title/Abstract])) OR (MODY, type 3[Title/Abstract])

# #3 #1 OR #2

# #4 "hnf1a protein human"[Supplementary Concept] OR "Hepatocyte Nuclear Factor 1-alpha"[MeSH Terms]

# #5 (((((Hepatocyte Nuclear Factor 1-alpha[Title/Abstract]) OR (HNF1A[Title/Abstract])) OR (HNF1-alpha[Title/Abstract])) OR (nuclear protein LF-B1[Title/Abstract])) OR (LF-B1 transcription factor[Title/Abstract])) OR (HNF1 homeobox A protein[Title/Abstract])

# #6 #4 OR #5

# #7 "Diabetes Mellitus"[Mesh]

# #8 diabetes[Title/Abstract]

# #9 #7 OR #8

# #10 #6 AND #9

# #11 #3 OR #10

# Supplementary Tables

**Supplementary Table 1.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Articles | Year | Country | Sex | Age of diagnosis (years) | BMI(kg/m2) | Family history | HbA1c (%) | FPG (mmol/L) | PPG (mmol/L) | FC (ng/ml) | PC (ng/ml) | TG(mmol/L) | TC(mmol/L) | HDL-c(mmol/L) | LDL-c(mmol/L) | Complications | Treatment | Mutation |
| Xiaoyan, Ren, et al(1) | 2021 | China | male | 12 | 17.7 | yes |  | 7.4 | 18.5 | 1.32 | 2.76 | 0.83 | 5.08 | 1.14 | 2.61 | None | OHA | c.G392T/p.R131L |
| female | 24 |  | yes |  | 8 |  |  |  |  |  |  |  |  | Lifestyles |
| Mengyan, Xu, et al(2) | 2018 | China | Female | 35 | 20.83 | yes | 6.5 | 5.2 | 13.6 | 2.09 | 3.85 | 0.42 | 4.08 | 1.60 | 2.29 | none | OHA | c.512G>A/p.Arg171Gln |
| Female | 17 | 19.53 | yes | 10.2 | 8.5 | 20.4 | 1.75 | 4.04 | 1.15 | 3.98 | 1.28 | 2.98 | none | OHA | c.391C>T/p.Arg131Trp |
| Male | 35 |  | yes |  |  |  |  |  |  |  |  |  | DR | OHA | c.512G>A/p.Arg131Gln |
| Male | 37 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | c.391C>T/p.Arg131Trp |
| Male | 40 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| Male | 24 |  | yes |  |  |  |  |  |  |  |  |  | DR, DKD | OHA+INS |
| Xiaoli Kang, et al(3) | 2017 | China | female | 13 | 21.8 | yes | 10.8 | 16.82 | 28.39 | 1.10 | 2.71 |  |  |  |  | none |  | c.51C>G/p.Leu17Leu |
| female | 23 |  | yes |  |  | 12.6 |  |  |  |  |  |  |  | OHA | c.79A>C/p.Ile27Leu |
| female | 48 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | c.814C>A/p.Arg272Ser |
| female | 26 |  | yes |  |  |  |  |  |  |  |  |  | DN, DR | OHA | c.1375C>T/p.Leu459Leu |
| female | 14 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | c.1460G>A/p.Ser487Asn |
| female | 11 |  | yes | 10.8 |  |  |  |  |  |  |  |  | DR | INS | c.326+91A>G/IVSInt+91A>G |
| male | 16 |  | yes |  |  | 18.2 |  |  |  |  |  |  |  |  | c.1107+9C>G/IVS5nt+9C>G |
| female | 16 |  | yes | 10.1 | 16.82 | 23.21 |  |  |  |  |  | 3.38 |  |  | c.1501+7G>A/IVS7nt+7G>A |
| Liubov Desiatkina, et al(4) | 2015 | china | male | 19 | 26.85 | yes | 12.3 | 17.6 | 27 | 1.42 | 1.91 |  |  |  |  |  | OHA | c.1410G>A/p.S487N |
| female | 52 |  | yes | 8.3 | 8 | 12 |  |  |  |  |  |  |  | INS |
| Tianping Wang, et al(5) | 2014 | china | female | 17 | 18.7 | yes | 5.4 | 4.94 |  | 0.70 | 2.44 | 0.77 | 4.0 | 1.20 | 2.45 | DN | INS | c.1130-1131insC/p.V380cfs\*39 |
| female | 36 | 19.6 | yes | 6.3 | 6.11 | 10.2 | 0.92 | 3.61 | 1.11 | 2.60 | 0.47 | 1.625 | DN,DR | lifestyles |
| female | 24 | 17.3 | yes | 9.2 | 10.88 |  | 0.92 | 1.80 | 0.84 | 4.40 | 0.99 | 3.028 | DN,DR | OHA |
| Xin pan,et al(6) | 2021 | china | female | 22 |  |  | 7.5 | 9.79 | 23.03 | 2.32 | 4.36 | 0.8 | 3.84 | 0.93 | 2.59 | none | OHA | p.P379T |
| Yuecheng Zhang, et al(7) | 2015 | china | female | 14 | 16.3 | yes | 5.7 | 6.2 | 6.8 |  |  | 1.65 |  |  |  | none |  | c.293C>T/p.X466W |
| Yipaerguli．Ainiwaner, et al(8) | 2021 | china | male | 6 | 16.5 | yes | 6.5 | 6.13 | 12.83 | 0.6 | 2.45 |  |  |  |  | none | OHA | c.A1398G/p.X466W |
| female | 46 | 20.24 | yes | 10.8 | 13.7 | 18.52 | 1.43 |  |  |  |  |  | none | OHA+INS |
| male | 30 | 21.22 | yes | 7.2 | 7.72 | 10.39 | 2.2 |  |  |  |  |  | none | OHA |
| male | 75 | 24.22 | yes |  | 12.29 | 17.83 |  |  |  |  |  |  | Coronary heart disease | OHA+INS |
| female | 48 | 27.29 | yes | 6.9 | 5.58 | 11.68 |  |  |  |  |  |  | none | OHA+INS |
| Yanwen Shu, et al(9) | 2021 | china | female | 26 | 20.47 | no | 12 |  | 17.4 | 0.63 | 2.13 | 1.15 |  | 1.01 | 2.55 | DR, DKD, DN | INS | c.865dupC/p.G292Rfs\*25 |
| female | 21 | 26.31 | yes | 5.9 |  | 14 | 2.23 | 8.09 | 4.24 |  | 0.88 | 2.09 | none | OHA | c.961C>A/p.L108P |
| female | 12 | 19.74 | yes | 12.1 |  |  | 1.95 | 3.21 | 1.28 |  | 0.92 | 1.34 | none | OHA | c.323T>G/p.L108P |
| female | 43 | 18.8 | yes | 6.5 |  |  |  |  |  |  |  |  | none | OHA |
| Mingwei Shao, et al(10) | 2020 | china | male | 12 | 20.8 | yes | 9.3 | 7.4 | 17.3 |  |  |  |  | 1.49 |  |  | OHA | c.779C>T/p.Thr260Met |
| male | 38 | 22.39 | yes | 9.4 | 12.4 | 26.8 |  |  |  |  |  |  |  | OHA |
| Ying Zhang, et al(11) | 2021 | china | female | 19 | 23.1 | yes | 9.6 | 11.2 | 20.6 | 0.05 | 3.34 |  |  |  |  | DR, DN | OHA | c.G812A/p.R271Q |
| Nikolai Paul Pace, et al(12) | 2019 | Malta | female | 14 | 19.56 | yes | 9.9 |  |  | 2.1 |  | 1.95 | 4.44 | 0.82 | 2.73 |  | INS | c.872dup/p.Gly2992fs |
| female | 16 | 23.32 | yes | 7.0 | 5.67 | 14.21 | 1.6 |  | 1.17 | 3.08 | 0.40 | 2.15 |  | INS |
| female | 19 | 22.65 | yes | 8.3 | 8.64 | 17.24 | 2.8 |  | 0.63 | 4.57 | 1.91 | 2.37 |  | INS |
| Pedro J, et al(13) | 2011 | Spain | Male11/female 4 | 17.6±4.4 | 26.0±4.2 | 2 patients | 7.4±1.5 |  |  |  |  |  |  | 2.90±0.90 |  |  | OHA 6/INS 3/OHA+OMNS 2 |  |
| Fernando M.A. et al(14) | 2016 | Brazil | Male 11/female 20 | 24±12 | 22±4 | yes | 7.0±1.4 | 7.8±3.1 |  | 0.5±0.2 |  | 9.2±3.6 | 5.0±0.9 | 1.4±0.3 | 3.1±0.7 |  | OHA 28/INS 5 |  |
| Maja D. Jesic, et al(15) | 2008 | Serbia | female | 10 | 20 | yes | 7.9 | 8.7 | 16.2 |  |  | 1.70 | 3.96 | 0.72 | 2.47 | Peripheral arterial disease | OHA | c.368T>C/p.Leu123Pro |
| female | 14 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| Junling Fu, et al(16) | 2019 | china | Total 12 | 21.75±8.74 | 20.70±1.65 |  | 8.31±2.62 | 8.26±3.17 | 13.46±6.34 | 1.34±1.51 | 2.42±1.35 | 0.97±0.,43 | 4.41±0.93 | 1.32±0.39 | 2.57±0.86 |  |  |  |
| N. Tonooka, et al(17) | 2002 | japan | Male 6/female 12 | 11.6±1.7 | 20.1±3.0 | 1 patients |  |  |  |  |  |  |  |  |  | none | Lifestyles 4/OHA 5/INS 9 |  |
| W L Awa, et al(18) | 2011 | Austria | Male 16/female 28 | 14.1±5.8 |  | 21 patients | 6.8±1.2 |  |  |  |  |  |  |  |  |  | Lifestyles 9/INS 16/OHA+INS 10 |  |
| Birgit Knebel, et al(19) | 2016 | Germany | female | 43 | 27 | yes | 7.4 | 12.7 | 22.2 | 0.4 | 0.6 | 2.2 | 7.0 | 1.1 | 4.8 | none |  | c.1785C>T/p.Pro588Serc.1765-1766delCA |
| male | 39 | 30 | yes | 8.9 | 12.8 | 16.0 | 0.7 | 1.2 | 6.9 | 7.3 | 0.7 | 3.4 | none |  |
| H.C.Fehmann, et al(20) | 2003 | Germany | male | 16 |  | yes | 6.4 | 8.0 | 13.0 |  |  |  |  |  |  |  |  | c.224C>T/p.Peo224Ser |
| female | 25 |  | yes |  |  |  |  |  |  |  |  |  |
| S. Bacon, et al(21) | 2015 | England | Male 24/female 35 |  |  |  |  |  |  |  |  |  |  |  |  | DR 10/DKD 6/Coronary heart disease 4 | Lifestyles 15/ OHA 21/INS 15 |  |
| Aoife M Egan, et al(22) | 2015 | England | female | 12 |  | yes | 12.7 | 10 | 23 | 1.5 |  |  |  |  |  | DR, DKD | INS | c.391C>T/p.Arg131Trp |
| Rachel E J Besser, et al(23) | 2012 | England | female | 14 |  | yes | 9.3 | 9.3 |  |  |  |  |  |  |  | Coronary heart disease | OHA | c.608G>C/p.R203H |
| male | 30 | 30 | yes | 9.7 |  |  |  |  |  |  |  |  |  |  |  |
| Shivani Misra, et al(24) | 2020 | England | male | 15 | 27.8 | yes | 7.5 |  | 7.4 | 0.6 |  |  |  |  |  |  | INS | P.A251T/A251T |
| male | 12 | 28.7 | yes | 8.3 |  | 10.9 | 0.79 |  |  |  |  |  |  | INS |
| female | 16 | 23.2 | yes | 12.2 |  | 7 | 0.47 |  |  |  |  |  |  | INS |
| female | 29 | 25.4 | yes | 8.7 |  | 13.8 | 1.51 |  |  |  |  |  |  | OHA | p.A251T/N |
| Maricor K, et al(25) | 2013 | The USA | female | 10 | 29.6 | yes |  | 10.67 |  | 0.8 |  |  |  |  |  | DKD, DN | OHA+INS | c.598C>T/p.Arg200Trp |
| female | 9 | 30.8 | yes | 7.7 | 13.28 |  | 0.5 |  |  |  |  |  |  | INS |
| female | 10 | 32.5 | yes | 9.0 | 11.17 |  | 0.8 |  |  |  |  |  |  | INS |
| Yael Lebenthal, et al(26) | 2017 | England | female | 24 | 24 | no |  |  |  |  |  |  |  |  |  | DR | OHA |  |
| female | 16 | 22.5 | no |  |  |  |  |  |  |  |  |  | DR, DKD | OHA+INS |
| female | 11 | 21 | no | 7.9 |  |  |  |  |  |  |  |  |  | INS |
| female | 10 | 25.6 | no | 6.3 |  |  |  |  |  |  |  |  | DKD | INS |
| male | 21 | 21.8 | no |  |  |  |  |  |  |  |  |  | DR | OHA+INS |
| male | 50 | 19.5 | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| female | 23 | 27.3 | no |  |  |  |  |  |  |  |  |  | DR | OHA+INS |
| male | 22 | 21.5 | no | 7.8 |  |  |  |  |  |  |  |  |  | INS |
| male | 17 | 21.5 | no | 6.7 |  |  |  |  |  |  |  |  |  | INS |
| male | 14 | 23.1 | yes | 6.7 |  |  |  |  |  |  |  |  | DKD | OHA+INS |
| male | 45 | 22.7 | no |  |  |  |  |  |  |  |  |  |  | OHA |
| female | 49 | 21.9 | no | 7.1 |  |  |  |  |  |  |  |  |  | OHA |
| female | 10 | 27 | yes | 9.4 |  |  |  |  |  |  |  |  |  | INS |
| male | 13 | 24.2 | no | 8.1 |  |  |  |  |  |  |  |  |  | OHA+INS |
| female | 13 | 28.5 | yes | 8.9 |  |  |  |  |  |  |  |  |  | OHA+INS |
| Souhaïra Ben Khelifa, et al(27) | 2015 | Tunisia | female | 14 | 25.49 | yes | 10.8 | 11.23 |  | 0.52 |  | 1.03 | 4.92 | 1.24 |  | DKD | OHA | P291fsinsC |
| Magdalena Szopa, et al(28) | 2015 | Poland | Male 26/female 46 | 24.95±10.96 | 25.05±4.0 |  | 6.83±1.41 | 6.91±2.68 |  | 1.33±0.66 |  | 1.23±0.92 | 4.79±0.97 | 1.59±0.42 | 2.64±0.86 |  |  |  |
| K. A. Iwen, et al(29) | 2012 | Germany | female | 20 |  | yes | 6.3 | 4.5 | 16.9 |  |  |  |  |  |  |  | OHA | PA98V polymorphism + Q495X stop |
| Atsushi Iwabuchi, et al(30) | 2013 | Japan | male | 1 |  | yes | 7.8 | 7.7 | 18.7 |  |  |  |  |  |  |  | OHA | c.593delA/p.Lys198fs |
| B.Isomaa, et al(31) | 1998 | Finland | Male 29/female 28 | 26.7±2.9 | 24.6±4.5 |  | 7.3±1.6 |  |  |  |  | 1.53±0.82 | 5.3±1.0 | 1.40±0.21 |  | DR 18/DKD 19/DN 17/Coronary heart disease 9 | Lifestyles 19/OHA 17/INS 21 |  |
| M. Hummel, et al(32) | 2006 | Germany | female | 17 | 20.3 | yes | 7.2 |  |  | 1.2 |  |  |  |  |  | none | OHA+INS | c.653A>G/p.Tyr218Cys |
| Belgium | male | 18 | 21.9 | yes |  |  |  | 1 |  |  |  |  |  |  | INS |
| female | 15 | 18.1 | yes |  |  |  | 1.19 |  |  |  |  |  |  | INS |
| Itziar Estalella, et al(33) | 2007 | Spain | Total 24 | 12.7±4.6 |  |  | 7.6±2.3 | 11.3±3.2 |  |  |  |  |  |  |  |  | Lifestyles 4/OHA 1/INS 3 |  |
| Naieli Bonatto, et al(34) | 2012 | Brazil | female | 46 | 28.37 | yes | 6.4 | 9.17 | 10.33 | 2.52 |  |  |  |  |  | Microvascular and macrovascular complications  | INS |  |
| female | 36 | 30.30 | yes | 14.3 | 15.61 | 20.22 | 3.37 |  |  |  |  |  | none | INS |
| male | 26 | 26.47 | yes | 9.78 | 22.56 | 18.44 | 1.40 |  |  |  |  |  | none | INS |
| male | 38 | 33.57 | yes | 10.68 | 15.44 | 23.33 | 3.50 |  |  |  |  |  | none | OHA |
| female | 16 | 33.30 | yes | 11.3 | 14.44 | 22.11 | 1.20 |  |  |  |  |  | Microvascular and macrovascular complications  | INS |
| female | 40 | 28.58 | yes | 11.45 | 16.94 | 18.11 | 2.10 |  |  |  |  |  | macrovascular complications  | OHA |
| male | 45 | 31.94 | yes | 12.6 | 16.72 | 18 | 1.20 |  |  |  |  |  | Microvascular complications | OHA+INS |
| Wojciech Fendler, et al(35) | 2011 | poland | Male 26/female 11 |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 2/OHA 16/INS 19 |  |
| Chen Fang, et al(36) | 2015 | china | female | 19 | 22 | yes | 15 | 18 | 21.56 | 1.48 |  |  |  |  |  |  | OHA | R45X (CGA>TGA) |
| Nattachet Plengvidhya, et al(37) | 2019 | Thailand | female | 14 | 19.43 | yes | 6.7 | 7 |  |  |  | 2.97 | 8.43 | 3.84 |  | none | OHA | R203C |
| Letı´cia S, et al(38) | 2014 | Brazil | male | 23 | 24.7 | yes | 7.2 |  |  | 1.6 |  |  |  |  |  | DR, DKD | OHA | c.24-35dup12/p.Gln9-Leu12du |
| female | 27 | 19.2 | yes | 6.7 |  |  | 1.6 |  |  |  |  |  | none | OHA+INS | c.872dupC/p.Gly292fs |
| Xiaojing Wang, et al(39) | 2018 | china | Male 12/female 5 | 25.56±13.32 | 20.91±2.23 |  | 8.36±2.14 | 8.75±2.93 | 16.19±6.08 | 0.91±0.40 | 1.77±0.93 |  | 4.53±1.02 |  | 2.75±0.86 |  |  |  |
| Elizabeth B. Tatsi, et al(40) | 2019 | Greece | female | 13.5 | 21 | yes | 11.5 |  |  |  |  |  |  |  |  |  |  | c.1204A>T/p.Asn402Tyr |
| female | 12.5 | 22.8 | yes | 7.4 |  |  |  |  |  |  |  |  |  |  | c.599G>A/p.Arg200Gln |
| male | 30 |  | no |  |  |  |  |  |  |  |  |  |  |  |
| M. Szopa, et al(41) | 2015 | Poland | Male 47/female 16 | 23.5±9.8 | 23.9±3.8 |  | 7.0±1.4 | 7.3±2.8 |  | 1.3±0.7 |  | 1.2±0.9 | 4.8±0.9 | 1.6±0.4 | 2.6±0.8 | DR 15/DKD 36 | Lifestyles 4/OHA 35/INS 22 |  |
| J. Skupie, et al(42) | 2008 | Poland | Male 11/female 31 | 24.5±10.9 | 23.0±3.2 |  | 7.5±1.8 | 7.8±3.1 |  | 1.45±0.47 |  | 1.2±0.7 | 5.0±1.1 | 1.6±0.5 | 2.9±0.9 | DR 21/DKD 11/Coronary heart disease 4 | INS 23 |  |
| Ibrar Rafique, et al(43) | 2021 | Pakistan | male | 17 | 24.6 | yes | 8.2 |  |  |  |  |  |  |  |  |  |  | c.526+1G>A |
| Junnosuke Miura, et al(44) | 1997 | japan | female | 19 | 22 | yes | 14.0 | 13.83 |  |  |  |  |  |  |  |  | INS |  |
| Ariel Pablo Lopez, et al(45) | 2010 | Argentina | Total 8 | 20.86 | 23±1.5 |  | 7.5±1.6 | 11.39±2.24 |  |  |  |  |  |  |  |  | Lifestyles 0/OHA 8/INS 0 |  |
| Tim J. McDonald, et al(46) | 2012 | England | Male 41/female 226 |  | 24.9±4.6 |  | 7.5±1.6 |  |  |  |  |  | 4.9±1.3 | 1.46±0.48 |  |  |  |  |
| STEPANKA PRUHOVA, et al(47) | 2013 | Czech Republic | female | 4 | 20.1 |  | 15 |  |  |  |  |  |  |  |  |  | INS | p.Arg272His |
| male | 13 | 29.4 | yes | 13 | 34 |  |  |  |  |  |  |  |  | INS | p.Ser142Phe |
| TIINAMAIJA TUOMI, et al(48) | 2006 | Sweden | female | 23.9 | 25.0 |  | 5.4 | 4.3 |  |  |  |  |  |  |  |  | none |  |
| female | 44.9 | 22.4 |  | 6.6 | 7.0 |  |  |  |  |  |  |  |  | none |
| female | 30.8 | 22.6 |  | 8.1 | 10.8 |  |  |  |  |  |  |  |  | INS |
| male | 34.2 | 21.9 |  | 6.4 | 7.0 |  |  |  |  |  |  |  |  | OHA |
| male | 42.5 | 24.3 |  | 8.7 | 12.2 |  |  |  |  |  |  |  |  | OHA |
| female | 50.6 | 23.4 |  | 9.1 | 11.3 |  |  |  |  |  |  |  |  | OHA |
| female | 41.1 | 28.2 |  | 6.7 | 5.3 |  |  |  |  |  |  |  |  | none |
| female | 62.6 | 24.7 |  | 6.5 | 5.2 |  |  |  |  |  |  |  |  | OHA |
| male | 32.9 | 22.3 |  | 7.8 | 7.5 |  |  |  |  |  |  |  |  | none |
| male | 46.5 | 23.9 |  | 7 | 7.9 |  |  |  |  |  |  |  |  | none |
| male | 66.1 | 23.4 |  | 9.9 | 16.2 |  |  |  |  |  |  |  |  | OHA |
| female | 40.2 | 16.7 |  | 5.9 | 8.3 |  |  |  |  |  |  |  |  | OHA |
| female | 47.3 | 19.8 |  | 6.1 | 9.4 |  |  |  |  |  |  |  |  | OHA |
| female | 20.0 | 23.1 |  | 9 | 5.4 |  |  |  |  |  |  |  |  | OHA |
| female | 28.0 | 21.2 |  | 6.7 | 5.0 |  |  |  |  |  |  |  |  | none |
| A. K. Ovsyannikova, et al(49) | 2018 | Russia | female | 12 | 24.5 | yes | 7.7 |  |  | 1.01 |  |  |  |  | 3.4 | DR, DN |  | p.Ser6Arg |
| male | 21 | 22.4 | yes | 7.1 | 9 | 12.8 | 0.78 |  |  |  |  | 3.2 | none | OHA |
| female | 45 |  |  |  |  |  |  |  |  |  |  |  | DN | OHA |
| S Mongolu, et al(50) | 2009 | England | female | 27 |  | yes | 9.8 |  |  |  |  |  |  |  |  |  | OHA |  |
| Wolfgang J. Schnedl, et al(51) | 2020 | Austria | female | 18 | 23 | yes | 8.4 |  |  |  |  |  |  |  | 8.05 |  | OHA | c.815G>A/p.Arg272His |
| female | 41 | 22 | yes |  | 9.19 |  |  |  |  |  |  | 6 |  | OHA | c.675delC/p.Ser225Argfs\*8 |
| Giuseppina Salzano, et al(52) | 2019 | Italy | female | 10 |  | no | 10.8 | 21.4 |  | 1.07 | 2.52 |  |  |  |  | none | OHA | c.709A>G |
| Marianne Becker, et al(53) | 2014 | Germany | female | 12 | 23.7 | yes | 7.4 |  |  |  |  |  |  |  |  | none | OHA | c.872dupC/p.Gly292ArgfsX25 |
| male | 14 | 19.2 |  | 7.0 |  |  |  |  |  |  |  |  | none | OHA | c.162T>C/p.Leu54Pro |
| Abdelhadi M. Habeb, et al(54) | 2011 | England | male | 7 |  | yes | 7.8 | 7.7 | 18.7 |  |  |  |  |  |  |  | OHA | c.526+1G>A |
| Maltoni G, et al(55) | 2012 | Italy | male | 15 |  | yes | 7.2 | 6.33 | 14.22 | 1.9 |  |  |  |  |  |  | OHA | c.92G>A/p.G31D |
| Akie Nakamura, et al(56) | 2012 | japan | female | 9 | 15.9 |  | 7.3 | 5.78 | 18.17 |  |  | 7.68 | 11.19 | 2.43 |  |  | INS |  |
| MC Ng, et al(57) | 2000 | china | female | 19 | 19.6 | yes | 8.0 |  |  | 0.47 |  |  |  |  |  | DR,DKD | INS | IVS2nt-1G>A |
| female | 24 | 22.7 | yes | 6.4 |  |  | 0.86 |  |  |  |  |  | DR. DKD | INS |
| female | 38 | 18.5 | yes | 17.2 |  |  | 0.32 |  |  |  |  |  | DR, DKD, DN, Coronary heart disease | INS |
| female | 12 | 18.8 | yes | 8.8 |  |  | 0.13 |  |  |  |  |  |  | INS |
| male | 15 | 16.6 | yes | 5.3 |  |  | 0.36 |  |  |  |  |  |  | INS |
| Jose Bernardo Quintos, et al(58) | 2013 | The USA | female | 13 | 21 | yes | 8.5 | 10.72 |  |  |  | 4 | 9.95 | 1.89 | 7.24 |  | OHA | c.1129delC/p.Leu377fsX7 |
| MP Selwood, et al(59) | 2008 | England | female | 25 |  | yes |  |  |  | 0.57 |  |  |  |  |  | DR, DKD, DN |  |  |
| female | 37 | 26.2 | yes | 7.2 | 9.4 |  |  |  |  |  |  |  | none | OHA |  |
| male | 26 |  | yes | 6.4 | 8.0 | 13.0 |  |  |  |  |  |  | none | Lifestyles |  |
| NAOKO IWASAKI, et al(60) | 1998 | japan | Male 6/female 5 | 16.1±2.0 | 20.6±0.4 |  |  |  |  |  |  | 1.53±0.82 | 5.3±1.0 | 1.40±0.21 |  | DR 5/DKD 4 | Lifestyles 2/OHA 3/INS 6 |  |
| M.P. Kyithar, et al(61) | 2010 | Ireland | Male 13/female 18 | 25±2 | 24.52±0.69 |  | 7.21±0.22 | 7.34±0.43 |  | 2.02±0.36 |  | 0.91±0.12 | 4.34±0.19 | 1.42±0.09 | 2.56±0.15 |  | Lifestyles 0/OHA 13/INS 8 |  |
| ALESSANDRO DORIA, et al(62) | 1999 | Poland | Male 32/female 47 | 21±10 |  |  | 6.9±1.6 | 6.44±2.06 | 14.39±5.39 |  |  | 5.41±3.14 | 10.54±2.22 | 2.86±0.76 |  | DR 5/DKD 14/Coronary heart disease 14 | Lifestyles 24/OHA 18/INS 37 |  |
| Miao Zhang, et al(63) | 2015 | china | female | 36 | 19.6 | yes | 6.3 | 6.11 | 10.12 | 0.31 | 1.20 | 1.11 | 2.60 | 0.47 | 1.625 |  | OHA | p.379fsinsC |
| female | 24 | 17.3 | yes | 9.2 | 10.88 |  | 0.31 | 0.60 | 0.84 | 4.40 | 0.99 | 3.028 | DR, DN |  |
| female | 23 | 18.7 | yes | 5.4 | 7.76 | 19.84 | 0.24 | 0.69 | 0.77 | 4.00 | 1.20 | 2.45 | DR, DN | OHA |
| female | 11 | 19.6 | yes | 7.3 | 5.88 | 13.37 | 0.53 | 1.25 | 0.85 | 4.15 | 1.52 | 2.24 | DN |  |
| Nattachet Plengvidhya, et al(64) | 2009 | Thailand |  | 31 | 24.49 |  | 6.60 | 6.94 |  | 0.3 |  | 0.76 | 5.10 | 1.26 | 3.49 |  |  | P475L |
| 12 | 20.41 |  | 4.90 | 9.27 |  | 0.66 |  | 0.43 | 3.55 | 1.58 | 1.77 |  |  | G554SfsX556 |
| S. Pruhova, et al(65) | 2003 | Czech Republic | Male 3/female 4 |  |  |  |  |  |  |  |  |  |  |  |  | DR 1/DKD 1/DN 1 | Lifestyles 2/OHA 1/INS 4 |  |
| Juraj Stanik, et al(66) | 2014 | Czech Republic |  | 16 | 20.2 |  | 6.8 | 5.4 |  | 1.29 |  |  |  |  |  |  | INS | c.392G>A/R131Q |
|  | 14 | 22.9 |  | 7.3 | 12.0 |  | 0.59 |  |  |  |  |  |  | INS  |
|  | 15 | 18.0 |  | 5.9 | 5.4 |  | 0.79 |  |  |  |  |  |  | Lifestyles | c.392G>C/R131P |
|  | 11 | 19.9 |  | 6.2 | 6.2 |  | 1.41 |  |  |  |  |  |  | INS | c.436C>G/Q146E |
|  | 15 | 22.0 |  | 8.6 | 5.8 |  | 1.33 |  |  |  |  |  |  | INS | c.1256C>G/S419\* |
| MACIEJ T. MALECKI, et al(67) | 2005 | Poland | female | 17 |  | yes |  |  |  |  |  |  |  |  |  | DR, DKD, macrovascular complications, | INS | IVS7nt-6G>A |
| female |  |  | yes |  |  |  |  |  |  |  |  |  |  | INS | Arg271Trp |
| GAYA THANABALASINGHAM, et al(68) | 2012 | England | Male  | 13 | 23.4 | yes |  | 21.3 |  | 1`.08 |  |  |  |  |  |  |  | c.92G>A/G31D |
| female | 29 | 22.5 | no |  | 11.2 |  | 0.50 |  |  |  |  |  |  |  | c.1469T>G/M490R |
| Maraschin et al.(69) | 2008 | Poland | Male 2/female 8 | 21±5 | 24±3 | 10 patients | 7.06±1.6 | 6.9±0.9 |  |  |  | 4.38(2.22-7.03) | 10.6±2 | 3.14±0.4 |  | DR 4/DKD 2/DN 1/Coronary heart disease3 |  |  |
| Catherine Pihoker, et al(70) | 2013 | The USA | Male 7/female 19 | 12.2±3.0 | 26.7±6.8 | 16 patients | 6.9±1.7 |  |  | 2.3±1.2 |  |  |  |  |  |  | Lifestyles 2/OHA 17/INS 15 |  |
| Jian Yu Xu, et al(71) | 2005 | china | Male 9/female 12 | 21.24±4.71 | 22.59±3.45 | 21 patients | 7.47±1.17 | 6.7(5.4-10) |  | 0.26(0.20-0.48) | 1.74(1.16-2.87) | 0.8(0.63-1.2) | 4.72±0.79 | 1.4(1.1-1.63) | 2.84±0.62 | DR 4/DKD 1/ DN 1/Coronary heart disease 0 | Lifestyles 5/OHA 11/INS 5 |  |
| I. Yoshiuchi, et al(72) | 1999 | japan | female | 25 | 22.2 |  | 7.6 |  |  |  |  |  |  |  |  | DR | INS | G415R |
| male | 30 | 17.3 | yes |  |  |  |  |  |  |  |  |  | DR, DKD, DN | OHA | R272C |
| male | 29 |  | yes | 6.3 |  |  |  |  |  |  |  |  | none | OHA+INS | A site mutation +102G>C |
| R.D. Cox, et al(73) | 1999 | England |  | 42 | 25 | yes |  | 6.2 |  |  |  |  |  |  |  |  | Lifestyles | Promoter -237G>A/G237A |
|  | 33 | 22 | no |  | 7.2 |  |  |  |  |  |  |  |  | Lifestyles | Promoter -8-9 2bp deletion |
|  | 45 | 25 | no |  | 7.2 |  |  |  |  |  |  |  |  | INS | A301T |
|  | 37 | 23 | no |  | 8 |  |  |  |  |  |  |  |  | OHA | T492I |
|  | 46 | 38 | yes |  | 13 |  |  |  |  |  |  |  |  | OHA | S498R |
| S. Demol, et al(74) | 2014 | Israel | male | 13.5 | 32.6 |  | 6.5 | 5.94 |  |  |  |  |  |  |  |  | INS | c.618G>A/p.W206X |
| male | 30 | 26.6 |  | 5.1 | 5.44 |  |  |  |  |  |  |  |  | INS |
| female | 19 | 28 |  | 7.5 | 7.83 |  |  |  |  |  |  |  |  | INS |
| female | 10.5 | 33.6 |  | 11.1 | 7.78 |  |  |  |  |  |  |  |  | INS |
| female |  | 21.1 |  | 5.1 | 5.17 |  |  |  |  |  |  |  |  | none |
| male |  | 28.3 |  | 5.3 | 5.44 |  |  |  |  |  |  |  |  | none |
| female |  | 20.1 |  | 4.8 | 5.22 |  |  |  |  |  |  |  |  | none |
| male |  | 20.8 |  | 5.4 | 5.5 |  |  |  |  |  |  |  |  | none |
| Aarón Domínguez-López, et al(75) | 2005 | Mexico | female | 12 | 27.3 | yes |  |  | 22.2 |  |  |  |  |  |  | none | none |  |
| Timothy M. Frayling, et al(76) | 2001 | England | Total 1 | 21 |  | no |  |  |  |  |  |  |  |  |  |  | OHA 1 | P129T |
| Total 3 | 18.7(9-31) |  | yes |  |  |  |  |  |  |  |  |  |  | OHA 2/INS 1 | R131W |
| Total 2 | 15.5(11-20) |  | yes |  |  |  |  |  |  |  |  |  |  | OHA 1/INS1 | R159W |
| Total10 | 20.4(14-38) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 3/OHA3/ INS 4 | P291fsinsC |
| Total 4 | 27.5)12-70) |  | yes |  |  |  |  |  |  |  |  |  |  | OHA 1/INS 3 |
| Total 3 | 17(14-23) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 1/INS 1 |
| Total 1 | 21 |  | no |  |  |  |  |  |  |  |  |  |  | OHA 1 |
| Total 7 | 27.4(13-37) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 3/OHA 2/INS 2 |
| Total 3 | 22(8-45) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 2 |
| Total 2 | 23.5()21-26) |  | yes |  |  |  |  |  |  |  |  |  |  | OHA 2 |
| Total 5 | 16.8(12-21) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 3/OHA 2 | P379fsdelCT |
| Total 2 | 19(13-25) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/INS 1 |
| Total 4 | 21.5(10-30) |  | yes |  |  |  |  |  |  |  |  |  |  | OHA 3/INS 1 | A443fsdekCA |
| Total 7 | 20.3(6-36) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 2/INS 4 | P447L |
| Total 3 | 17(17-18) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 2/INS 1 | P159L |
| Total 7 | 34(15,77) |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 5/INS 1 | T620I |
| François Godart, et al(77) | 2000 | The USA | male | 26 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | >35nt -218T>C |
| male | 19 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | >35NT -207-206ins27 |
| male | 20 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles | 15nt -187C>T |
| male | 4 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles | 12nt -119delG |
| female | 29 |  | yes |  |  |  |  |  |  |  |  |  |  | INS | 12nt -119G>A |
| female | 32 |  | yes |  |  |  |  |  |  |  |  |  |  | INS | -97T>G |
| male | 23 |  | yes |  |  |  |  |  |  |  |  |  |  | INS | -62C>G |
| Torben Hansen, et al(78) | 1997 | The USA | female | 16 | 21.2 | yes | 8.2 | 12.5 |  |  |  |  |  |  |  |  |  | Exon2 128 ATC(Ile)>AAC（Asn) |
| female | 13 | 21.4 | yes | 6.4 | 7.2 |  |  |  |  |  |  |  |  |  | Exon6 379 CCT(Pro)>CC (frameshift) |
| male | 18 | 22.1 | yes | 7.2 | 7.7 |  |  |  |  |  |  |  |  |  | Exon7 447 CCG(Pro)>CTG(Leu) |
| male | 7 | 21.5 | yes | 9.1 | 14.8 |  |  |  |  |  |  |  |  |  | Exon2 143 CAC(His)>TAC(Tyr) |
| male | 15 | 25.0 | yes | 9.2 | 12.1 |  |  |  |  |  |  |  |  |  |  |
| female | 12 | 33.7 | yes | 6.3 | 6.9 |  |  |  |  |  |  |  |  |  |  |
| female | 5 | 23.2 | yes | 6.0 | 7.0 |  |  |  |  |  |  |  |  |  |  |
| female | 17 | 40.2 | yes | 12.7 | 12.7 |  |  |  |  |  |  |  |  |  |  |
| female | 19 | 22.9 | yes | 6.1 | 6.1 |  |  |  |  |  |  |  |  |  | Exon9 559 GCA(Ala)>GCAA(frameshift) |
| Y. Horikawa, et al(79) | 2014 | japan | Male 6/female 18 | 11.9±2.9 | 19.5±2.8 | 22 patients  |  | 7.1±1.2 |  | 1.5±0.5 |  |  |  |  |  |  | Lifestyles 4/OHA 10/INS 10 |  |
| T. Ikema, et al(80) | 2002 | japan | male | 1 |  |  |  |  |  |  |  |  |  |  |  | DR, DKD | INS | L518P519fsTCC>A+T521L+V617I |
| female | 47 | 23.4 |  |  |  |  |  |  |  |  |  |  |  | Lifestyles |  |
| male |  | 17.5 |  |  |  |  |  |  |  |  |  |  |  |  | L518P519fsTCC>A |
| female | 13 | 19.0 |  |  |  |  |  |  |  |  |  |  |  | INS | T512L+V617L |
| female | 10 | 20.9 |  |  |  |  |  |  |  |  |  |  |  | OHA | Primer A+102G>C |
| male | 38 | 23.5 |  |  |  |  |  |  |  |  |  |  |  | OHA |  |
| Timothy M, et al(81) | 1997 | England | Total 44 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 5/OHA 23/INS 10/OHA+INS 1 | P291fsinsC |
| Total 6 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | OHA 5/INS 1 | P291fsdelC |
| Total 5 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 4 | W267X |
| Total 3 patients  |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 2 | E132K |
| Total 3 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | OHA 1/INS 2 | IVS2nt+1G>A |
| Total 5 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | INS 5 | IVS4nt-2A>G |
| Total 2 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | INS 2 | D135fsdelA |
| Total 3 patients  |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 2 | A443fsdelCA |
| 1 patient | 29 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles  | Q474X |
| Total 2 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 2 | IVS8nt+1G>A |
| Total 3 patients |  |  |  |  |  |  |  |  |  |  |  |  |  | OHA 2/INS 1 | IVS9nt-1G>A |
| Maria Galán, et al(82) | 2010 | Spain | female | 20 | 21 | yes | 5.9 | 6.9 |  |  |  |  |  |  |  |  | Lifestyles | c.55G>C/exon1 delCACGCGG |
| male | 17 | 24 | yes | 7.4 | 10.2 |  |  |  |  |  |  |  |  | OHA | c.397G>A/p.Val133Met |
| female | 24 | 20 | yes | 9.6 | 7.9 |  | 1.9 |  |  |  |  |  |  | OHA | c.586A>G/p.Thr196Ala |
| female | 14 | 22 | yes | 10.6 | 14.2 |  | 1.0 |  |  |  |  |  |  | OHA+INS | c.704A>G/p.Glu235Gly |
| female | 17 | 22 | yes | 5.8 | 4.7 |  | 0.8 |  |  |  |  |  |  | OHA | c.811C>T/p.Arg271Trp |
| male | 34 | 25 | yes | 5.8 | 7.0 |  | 0.76 |  |  |  |  |  |  | Lifestyles | c.1136C>G/p.Pro379Arg |
| A. Johansen, et al(83) | 2005 | Denmark | Male 19/female 21 | 24.0±12.1 | 25.4±4.4 | 20 patients yes | 7.8±1.4 |  |  | 1.06±0.57 |  | 1.51±1.58 | 5.3±1.3 | 1.48±0.42 |  | DR 2/DKD 2/DN 1 | Lifestyles 2/OHA 7/INS 10 |  |
| K.-A. Kim, et al(84) | 2003 | Korea | male | 15 | 18.3 | yes |  | 5.1 | 18.8 | 0.30 | 0.99 |  |  |  |  |  | OHA+INS | R263L |
| Naoko Iwasaki, et al(85) | 1997 | The USA | male | 64 | 20.9 | yes |  | 6.2 |  |  |  |  |  |  |  | none | Lifestyles | G191D |
| male | 17 | 17.5 | yes |  | 5.1 |  |  |  |  |  |  |  | none | Lifestyles | K205Q |
| female | 15 | 19.5 | yes |  | 10.2 |  |  |  |  |  |  |  | none | OHA | P379fsdelCT |
| male | 13 | 20.3 | yes |  |  |  |  |  |  |  |  |  | DR, DKD | INS | L584S585fsinsTC |
| male | 28 | 22.2 | yes |  | 7.4 |  |  |  |  |  |  |  | none | Lifestyles | R263C |
| female | 16 | 22.4 | yes |  |  |  |  |  |  |  |  |  | DR | INS | T392fsdelA |
| female | 9 | 20.9 | yes |  |  |  |  |  |  |  |  |  | DR, DKD | INS | R131Q |
| male | 9 | 20.7 | yes |  | 6.1 |  |  |  |  |  |  |  | none | Lifestyles | L12H |
| T-S. Jap, et al(86)  | 2000 | china | male | 23 | 19.81 | yes | 7.4 |  |  |  |  | 1.02 | 4.19 |  |  | DR | OHA |  |
| Henian Cao, et al(87) | 2002 | Canada | female | 19 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | c.105delCA/1051delCA |
| female | 13 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| male | 12 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles |
| female | 16 |  | yes |  |  |  |  |  |  |  |  |  |  | INS | c.415G>A/R131Q |
| female | 13 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| male | 38 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | c.771C>T/Q250X |
| male | 23 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles |
| female | 15 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA |
| Soo Heon Kwak, et al(88) | 2016 | Korea | female | 19 | 21.3 | yes | 6.1 | 6.67 | 0.9 |  |  |  |  |  |  |  | OHA | c.415C>G/p.Leu139Val |
| Doddabelavangala Mruthyunjaya M, et al(89) | 2017 | India | female | 22 | 30 | yes |  |  |  |  |  |  |  |  |  |  | INS | c.8C>G/Ser3Cys |
| E. Stern, et al(90) | 2007 | Israel | male | 13 |  | yes | 11 | 9.94 |  |  |  |  |  |  |  |  | INS | Arg131Gln |
| female | 14.5 |  | yes | 10.4 |  |  |  |  |  |  |  |  |  | INS | Arg159Trp |
| male | 16 |  | yes | 7.1 | 6.56 |  |  |  |  |  |  |  |  | Lifestyles | Ala174fsdelCGCAGCGTAAG |
| male | 18 |  | yes | 4.6 | 15.56 |  |  |  |  |  |  |  |  | INS | Arg271Gln |
| male | 15 |  | yes | 6.1 | 6.56 |  |  |  |  |  |  |  |  | Lifestyles | Pro447Leu |
| male | 13 |  | yes | 11.8 | 23.17 |  |  |  |  |  |  |  |  | Lifestyles | Pro291fsinsC |
| Yorifuji T, et al(91) | 2012 | japan | female | 15 |  |  |  | 9.17 |  |  |  |  |  |  |  |  |  | c.1043T>C/p.L348P |
| female | 12 |  |  |  | 6.88 |  |  |  |  |  |  |  |  |  | c.779C>T/p.T260M |
| male | 9 |  |  |  | 6.88 |  |  |  |  |  |  |  |  |  | c.391C>T/p.R131W |
| male | 11 |  |  |  | 9.16 |  |  |  |  |  |  |  |  |  | Exon 7-9 deletion |
| male | 8 |  |  |  | 4.66 |  |  |  |  |  |  |  |  |  | c.788G>A/p.R263H |
| male | 13 |  |  |  | 4.61 |  |  |  |  |  |  |  |  |  | c.872delC/p.P291fs |
| female | 13 |  |  |  | 7.6 |  |  |  |  |  |  |  |  |  | c.1181delC/p.P394fs |
| female | 11 |  |  |  | 8.49 |  |  |  |  |  |  |  |  |  | c.1054delT/p.S352fs |
| female | 11 |  |  |  | 9.27 |  |  |  |  |  |  |  |  |  | c.392G>A/p.R131Q |
| female | 13 |  |  |  | 5.44 |  |  |  |  |  |  |  |  |  | c.872-873insC/p.P291fs |
| male | 14 |  |  |  | 4.27 |  |  |  |  |  |  |  |  |  | c.598C>T/p.R200W |
| S. Tanaka, et al(92) | 2000 | japan |  | 27 | 21.3 | yes | 6.4 | 10.9 |  |  |  |  |  |  |  |  | Lifestyles | T539fsdelC |
|  | 32 | 21.4 | yes |  | 7.3 |  |  |  |  |  |  |  |  | Lifestyles |
|  | 27 | 18.5 | yes |  | 7.5 |  |  |  |  |  |  |  |  |  | P291fsinsC |
|  | 33 | 18.7 | yes |  | 8.4 |  |  |  |  |  |  |  |  |  |
| Dalia Toaima, et al(93) | 2005 | Germany |  | 14 | 22.8 | yes | 6.8 | 8.0 |  | 0.97 |  |  |  |  |  |  | OHA | c.73G>C/A25P |
|  | 10 | 25.8 | yes | 7.1 | 8.8 |  | 2.3 |  |  |  |  |  |  | OHA | c.499G>A/R200Q |
|  | 15 | 21.7 | yes | 6.3 | 8.2 |  |  |  |  |  |  |  |  | INS | c.873-874insC/P291fsinsC |
|  | 16 | 22.0 | yes | 6.4 | 9.4 |  | 0.79 |  |  |  |  |  |  | INS | c.1155-1156insA/L383fsinsA |
|  | 11 | 20.2 | yes | 8.7 | 13.1 |  | 0.5 |  |  |  |  |  |  | INS | c.1323delG/R442fsX456 |
| Maurizio Delvecchio, et al(94) | 2014 | Italy | Children 6 | 14.1-17.8 |  |  | 5.9-8.1 | 6.06-8.28 |  |  |  |  |  |  |  |  | Lifestyles 3/OHA 1/INS 2 |  |
| Adults 3 | 17.9-19.6 |  |  | 7,2-9.5 | 6.89-12.17 |  |  |  |  |  |  |  |  | Lifestyles 1/INS 2 |  |
| Christina Tatsia,et al(95) | 2013 | Greece |  | 18 |  |  |  | 8.33 |  |  |  |  |  |  |  |  |  | C.319C>T/R131W |
|  | 14 |  |  |  | 8.89 |  |  |  |  |  |  |  |  |  | c.481G>A/A161T |
|  | 14 |  |  |  | 12.22 |  |  |  |  |  |  |  |  |  | c.481G>C/A161P |
|  | 8 |  |  |  | 8.5 |  |  |  |  |  |  |  |  |  | c.493T>C/W165R |
|  | 0.3 |  |  |  | 14.2 |  |  |  |  |  |  |  |  |  | c.599G>A/R200Q |
|  | 17 |  |  |  | 9.44 |  |  |  |  |  |  |  |  |  | c.607C>T/R203C |
|  | 16 |  |  |  |  |  |  |  |  |  |  |  |  |  | c.682-684delGAG/E228del |
|  | 8 |  |  |  | 16.67 |  |  |  |  |  |  |  |  |  | c.685C>T/R229X |
|  | 13.5 |  |  |  | 14.11 |  |  |  |  |  |  |  |  |  |
|  | 24 |  |  |  | 5.56 |  |  |  |  |  |  |  |  |  | c.788G>A/R263H |
|  | 20 |  |  |  | 8.17 |  |  |  |  |  |  |  |  |  | c.824-826delAAG/E275del |
|  | 18 |  |  |  | 11.11 |  |  |  |  |  |  |  |  |  | c.872dupC/G292fs |
|  | 23 |  |  |  | 6.94 |  |  |  |  |  |  |  |  |  | c.1177delT/S393fs |
|  | 13 |  |  |  | 11.61 |  |  |  |  |  |  |  |  |  | c.1201C>T/Q401X |
|  | 3 |  |  |  | 10.5 |  |  |  |  |  |  |  |  |  | c.1331-1332delAG/S445fs |
|  | 20 |  |  |  | 9.11 |  |  |  |  |  |  |  |  |  | c.1501G>A/A501T |
| BEATE KARGES, et al(96) | 2007 | Germany | female | 13 | 25 | yes | 8.5 | 9.3 | 13.4 | 0.9 |  |  |  |  |  |  | OHA | c.526+1delGTAA |
| female | 14 |  | yes | 6.0 | 7.3 | 10.1 | 1,8 |  |  |  |  |  |  |  |
| female | 19 |  | yes | 6.1 | 6.8 | 13.1 | 1.7 |  |  |  |  |  |  |  |
| Atsushi Iwabuchi, et al(97) | 2013 | japan | male | 1.6 | 15.3 | yes | 7.9 | 5.1 | 19.78 | 0.2 |  |  |  |  |  |  | INS | c.593delA/p.Lys198fs |
| H. U. Irgens, et al(98) | 2013 | Norway |  | 9 | 28.3 | yes | 7.2 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.686G>A/p.Arg229Gln |
|  | 9 | 19.4 | yes | 6.8 |  |  |  |  |  |  |  |  |  | OHA | c.1136-1137delCT/p.Pro379fs |
|  | 2 | 17.6 | no | 4.7 |  |  | 1.5 |  |  |  |  |  |  | Lifestyles  | c.872dupC/p.Gly292fs |
|  | 13 |  | no | 7.9 |  |  | 0.4 |  |  |  |  |  |  |  |
|  | 13 | 18.1 | no | 8.5 |  |  |  |  |  |  |  |  |  | INS | c.686G>A/p.Arg229Gln |
|  | 9 | 18.3 | no | 8.7 |  |  | 0.9 |  |  |  |  |  |  | OHA | c.391C>T/p.Arg131Trp |
|  | 10 | 25.3 | yes | 8.6 |  |  | 1.0 |  |  |  |  |  |  | Lifestyles  | c.686G>A/p.Arg229Gln |
|  | 12 | 28.9 | yes | 7.4 |  |  | 1.0 |  |  |  |  |  |  | Lifestyles  | c.1745A>G/p.His582Arg |
|  | 14 | 18.6 | yes | 6.9 |  |  | 1.5 |  |  |  |  |  |  | OHA | c.872dupC/p.Gly292fs |
|  | 7 | 22.8 | yes | 6.8 |  |  | 0.45 |  |  |  |  |  |  | Lifestyles  | c.956-2A>G |
|  | 13 | 21.9 | no | 8.3 |  |  | 0.88 |  |  |  |  |  |  | Lifestyles  | c.1351A>G/p.Ser451Gly |
|  | 13 | 23.5 | yes | 10.4 |  |  | 0.66 |  |  |  |  |  |  | INS | c.666-668delGAA/p.Lys222del |
|  | 13 | 22.6 | yes | 7.1 |  |  | 0.78 |  |  |  |  |  |  | Lifestyles  |
|  | 4 | 16.6 | no |  |  |  |  |  |  |  |  |  |  |  | c.872dupC/p.Gly292fs |
|  | 7 | 22.1 | yes | 5.8 |  |  | 1.24 |  |  |  |  |  |  | Lifestyles  |
| S. Buchbinder, et al(99) | 2010 | Germany |  | 43 | 26 |  |  |  |  |  |  |  |  |  |  | none | OHA+INS | c.121G>T/p.E41X |
|  | 29 | 28 |  |  |  |  |  |  |  |  |  |  | DR, DN | OHA+INS |
|  | 30 | 24 |  |  |  |  |  |  |  |  |  |  | none | OHA+INS |
|  | 35 | 26 |  |  |  |  |  |  |  |  |  |  | none | Lifestyles  |
|  | 18 | 26 |  |  |  |  |  |  |  |  |  |  | none | OHA |
|  | 63 | 26 |  |  |  |  |  |  |  |  |  |  | none | OHA |
|  | 38 | 22 |  |  |  |  |  |  |  |  |  |  | none | OHA |
|  | 53 | 29 |  |  |  |  |  |  |  |  |  |  | none | OHA |
|  | 21 | 29 |  |  |  |  |  |  |  |  |  |  | none | INS |
|  | 34 | 28 |  |  |  |  |  |  |  |  |  |  | none | Lifestyles  |
| Z. Bazalova, et al(100) | 2010 | Czech Republic | male | 24 | 23.8 | yes | 5.0 | 7 |  | 2.21 |  | 0.56 | 3.50 | 1.04 | 2.21 | none | Lifestyles  | c.802T>C/p.Phe268Leu |
| male | 47 | 28.0 | yes | 5.6 | 7.5 |  | 1.97 |  | 0.95 | 6.06 | 1.70 | 3.93 | none | Lifestyles  |
| male | 42 | 34 | yes | 5.3 | 6.5 |  | 1.96 |  | 1.03 | 4.49 | 0.98 |  | none | OHA |
| male | 27 | 24.7 | yes | 7.7 | 8 |  | 0.90 |  | 0.83 | 5.19 |  |  | DR, DN | INS | c.871C>T/p.Pro291Ser |
| male | 27 | 24.5 | yes | 3.9 | 8.5 |  | 1.11 |  | 2.06 | 4.69 | 1.38 | 2.55 | none | Lifestyles  |
| R. BARRIO, et al(101) | 2002 | Spain | Male 2/female 3 | 14.4±2.7 |  |  | 6.7±1.5 | 8.09±1.91 | 15.95±2.43 |  |  |  |  |  |  | DKD 3 | Lifestyles 1/OHA 2/INS 2 |  |
| G. Alkorta-Aranburu, et al(102) | 2014 | The USA | male | 10 |  |  |  |  |  |  |  |  |  |  |  |  | INS | c.391C>T/p.Arg131Trp |
| male | 3 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles  | c.586A>G/p.Thr196Ala |
| Suwattanee Kooptiwut, et al(103) | 2009 | Thailand | male | 12 | 20/41 | yes |  | 13.2 |  |  |  | 0.43 | 3.55 |  | 1.77 |  | OHA | G554fsX556 |
| female | 50 | 22.51 | yes |  | 11.71 |  |  |  | 0.723 | 4.82 |  | 3.24 | DR, DN | OHA |
|  | male | 52 | 18.34 | yes |  | 6.33 |  |  |  |  |  |  |  |  |  |
| Bente B. Johansson,et al(104) | 2016 | Norway |  | 6 | 15.5 | no | 10.8 |  |  |  |  |  |  |  |  |  | INS | c.66C>G/p.Ser22Arg |
|  | 14 | 15.8 | no | 11.4 |  |  |  |  |  |  |  |  |  | INS | c.1061C>T/p.Thr354Met |
|  | 13 | 24.4 | no | 6.2 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.335C>T/p.PRO112Leu |
|  | 13 | 24.1 | no | 6.6 |  |  |  |  |  |  |  |  |  |  |
|  | 9 | 18.3 | no | 8.7 |  |  |  |  |  |  |  |  |  | OHA | c.391C>T/p.Arg131Trp |
|  | 10 |  | yes | 10.6 |  |  |  |  |  |  |  |  |  | INS | c.523C>G/p.Gln175Glu |
|  | 14 | 26.3 | no |  |  |  |  |  |  |  |  |  |  | Lifestyles  | c.608G>A/p.Arg203His |
|  | 13 | 27.8 | no |  |  |  |  |  |  |  |  |  |  | Lifestyles  |
|  | 13 | 23.5 | yes | 10.4 |  |  |  |  |  |  |  |  |  | INS | c.666-668del/p.Lys222del |
|  | 13 | 22.6 | yes | 7.1 |  |  |  |  |  |  |  |  |  | Lifestyles  |
|  | 10 | 25.3 | yes | 8.6 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.686G>A/p.Arg229Gln |
|  | 15 | 27.3 | yes | 7.0 |  |  |  |  |  |  |  |  |  |  |
|  | 7 | 22.1 | yes | 5.8 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.872dup/p.Gly292ArgfsTer25 |
|  | 2 | 17.6 | no | 4.7 |  |  |  |  |  |  |  |  |  | Lifestyles  |
|  | 6 |  | no | 9.2 |  |  |  |  |  |  |  |  |  | INS | c.917G>T/p,Gly306Val |
|  | 12 |  | no |  |  |  |  |  |  |  |  |  |  |  | c.1061C>T/p.Thr354Met |
|  | 13 | 21.9 | no | 8.3 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.1351A>G/p.Ser451Gly |
|  | 11 | 27.1 | no | 7.0 |  |  |  |  |  |  |  |  |  | INS | c.1640-1641del/p.Thr547ArgfsTer5 |
|  | 8 | 14.2 | no | 9.7 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.1739C>T/p.Pro580Leu |
|  | 12 | 28.9 | yes | 7.4 |  |  |  |  |  |  |  |  |  | Lifestyles  | c.1745A>G/p.His582Arg |
| Alessia Cappelli, et al(105) | 2009 | Italy | female | 17 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | R159W |
| male | 13 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | R272H |
| female | 16 |  |  |  |  |  |  |  |  |  |  |  |  | OHA+INS |
| female | 9 |  |  |  |  |  |  |  |  |  |  |  |  | OHA |
| female | 23 |  |  |  |  |  |  |  |  |  |  |  |  | INS |
| male | 16 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | IVS4nt-1G>T |
| male | 30 |  |  |  |  |  |  |  |  |  |  |  |  | OHA |
| A Costa, et al(106) | 2000 | Spain |  Ale 7/female 12 | 20.5±6.4 | 23.1±3.4 |  | 7.9±2.4 |  |  |  |  | 1.0±0.4 | 4.9±0.8 |  |  |  | Lifestyles 5.OHA 5/INS 9 |  |
| Klara Rozenkova, et al(107) | 2015 | Czech Republic | male | 6 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | p.Gly31Asp |
| male | 2.9 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles  | p.Asn62Lysfs93\* |
| male | 2.1 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles  | p.Leu254Gln |
| male | 46.3 |  | yes |  |  |  |  |  |  |  |  |  |  | Lifestyles  | p.Arg272His |
| female | 4.0 |  | yes |  |  |  |  |  |  |  |  |  |  | OHA | p.Glu508Lys |
| M. Lehto, et al(108) | 1999 | Sweden | Male 5/female 4 | 24±10 | 23.1±3.4 |  | 7.2±1.2 | 8.5±2.7 |  | 0.22±0.22 |  | 1.35±0.47 | 5.72±1.27 | 1.41±0.36 |  |  | Lifestyles 1/OHA 4/INS 4 |   |
| LISE BJØRKHAUG, et al(109) | 2003 | Norway | 1 | 12 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles  | G47E |
| 7 | 14-31 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 2/INS 1 | P112L |
| 11 | 10-20 |  |  |  |  |  |  |  |  |  |  |  | DR 1 | Lifestyles 1/OHA 4/INS 5 | R131W |
| 6 | 11-26 |  |  |  |  |  |  |  |  |  |  |  | DR 2/DN 1 |  | R171X |
| 1 | 31 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | T196fsdelCCAA |
| 4 | 21-36 |  |  |  |  |  |  |  |  |  |  |  | DKD 1 | INS 3 | R229Q |
| 1 | 24 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | IVS3-G>A |
| 1 |  |  |  |  |  |  |  |  |  |  |  |  |  |  | S256T |
| 6 | 13-27 |  |  |  |  |  |  |  |  |  |  |  |  |  | R263C |
| 1 | 16 |  |  |  |  |  |  |  |  |  |  |  |  | OHA | R271W |
| 1 | 24 |  |  |  |  |  |  |  |  |  |  |  | DKD 1 | Lifestyles  | A276D |
| 31 | 6-54 |  |  |  |  |  |  |  |  |  |  |  | DR 5/DKD 1/DN 3 | Lifestyles 4/OHA 10/INS 14 | P291fsinsC |
| 2 | 11-20 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 1/OHA 1 | P379fsdelCT |
| 3 | 12-13 |  |  |  |  |  |  |  |  |  |  |  | DR | INS 2 | S445fsdelAG |
| 1 | 17 |  |  |  |  |  |  |  |  |  |  |  |  | Lifestyles 1  | P447L |
| 1 |  |  |  |  |  |  |  |  |  |  |  |  |  |  | Q446X |
| 1 | 35 |  |  |  |  |  |  |  |  |  |  |  |  | OHA 1/INS 1 | S531T |

Abbreviations: BMI, body mass index; HbA1c, glycated hemoglobin; FPG, fasting plasma glucose; PPG, post-prandial plasma glucose; FC, fasting C-peptide; PC, post-prandial C-peptide; TG, triglyceride; TC, total cholesterol; HDL-c, high-density lipoprotein cholesterol; LDL-c, low-density lipoprotein cholesterol; DR, diabetic retinopathy; DKD,diabetic kidney disease; DN, diabetic neuropathy; OHA, oral hypoglycemic drugs; INS, insulin.

**Supplementary Table 2.** Characteristics of patients with *HNF1-alpha* MODY in different types of *HNF1-alpha* mutations.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Subject | Total (n = 492) | Missense mutations (n = 227) | Frameshift mutations (n = 198) | Nonsense mutations (n = 30) | Synonym mutations (n = 2) | Non-coding mutations (n=35) | P value |
| Age of diagnosis (years) | 20.1 (18.4,21.8) | 20.2 (17.3,23.1) | 20.2 (17.3,23.1) | 19.1 (16.8,21.5) | 19.5 (10.5,28.5) | 21.6 (17.7,25.5) | 0.881 |
| BMI (kg/m2) | 22.5 (20.3,24.7) | 23.3 (22.6,23.9) | 21.0 (20.3,21.6) | 25.8 (23.7,27.8) | NA | 20.5 (19.5,21.6) | < 0.001\* |
| HbA1c (%) | 7.9 (7.1,8.8) | 8.2 (7.9,8.5) | 7.3 (6.9,7.7) | 7.2 (5.9,8.5) | NA | 9.5 (8.0,11.0) | < 0.001\* |
| FPG (mmol/L) | 9.0 (8.5,9.6) | 9.3 (8.7,10.0) | 8.7 (8.2,9.2) | 8.6 (6.6,10.6) | NA | 10.1 (8.1,12.1) | 0.320 |
| 2h PG (mmol/L) | 17.5 (14.6,20.4) | 15.6 (14.7,16.4) | 14.5 (13.7,15.2) | 19.2 (18.2,20.3) | NA | 20.7 (19.7,21.8) | < 0.001\* |
| Fasting C-peptide (ng/mL) | 0.87 (0.55,1.19) | 1.17 (1.08,1.26) | 0.98 (0.83,1.13) | 0.89 (0.62,1.16) | NA | 0.43 (0.33,0.53) | < 0.001\* |
| 2-hour post-load C-peptide (ng/mL) | 2.42 (0.93,3.91) | 3.18 (2.91,3.45) | 1.66 (1.44,1.88) | NA | NA | NA | < 0.001\* |
| TG (mmol/L) | 1.50 (1.37,1.64) | 1.51 (1.36,1.66) | 1.45 (1.07,1.83) | NA | NA | NA | 0.772 |
| TC (mmol/L) | 4.81 (4.31,5.30) | 5.03 (4.82,5.24) | 4.52 (4.10,4.94) | NA | NA | NA | 0.034\* |
| HDL-c (mmol/L) | 1.23 (0.96,1.49) | 1.36 (1.25,1.47) | 1.09 (0.98,1.20) | NA | NA | NA | < 0.001\* |
| LDL-c (mmol/L) | 3.08 (2.84,3.33) | 3.18 (2.95,3.41) | 2.92 (2.57,3.27) | NA | NA | NA | 0.226 |
| Male (%) | 46.7 (39.9,53.7) | 49.6 (40.9,58.4) | 32.5 (19.9,48.3) | 50.0 (26.0,74.0) | 100 (0,100) | 50.0 (29.4,70.6) | 0.448 |
| Family history (%) | 46.9 (27.2,67.6) | 38.7 (13.9,71.2) | 33.3 (14.9,58.9) | 100 (0,100) | 100 (0,100) | 94.4 (69.4,99.2) | 0.048\* |
| Microvascular complications (%) | 40.7 (23.2,61.0) | 23.9 (15.2,35.5) | 45.5 (22.3,70.8) | 50.0 (16.8,83.2) | 50.0 (5.9,94.1) | 100 (0,100) | 0.389 |
| Diabetic retinopathy (%) | 33.3 (16.7,55.4) | 19.1 (11.2,30.6) | 31.1 (13.4,56.9) | 33.3 (8.4,73.2) | 50.0 (5.9,94.1) | 100 (0,100) | 0.699 |
| Diabetic kidney disease (%) | 17.7 (5.9,42.3) | 14.3 (7.3,26.1) | 9.8 (2.2,34.0) | NA | 0 (0,100) | 80.0 (30.9,97.3) | 0.046\* |
| Diabetic neuropathy (%) | 20.0 (10.2,35.5) | 13.5 (6.6,25.7) | 23.4 (6.3,58.2) | 16.7 (2.3,63.1) | 50.0 (5.9,94.1) | 20.0 (2.7,69.1) | 0.733 |
| Lifestyles (%) | 22.7 (18.7,27.2) | 22.9 (17.2,29.7) | 25.2 (15.9,37.4) | 15.0 (4.9,37.6) | 0 (0,100) | 52.3 (2.4,98.0) | 0.858 |
| OHA (%) | 43.0 (38.2,48.0) | 43.5 (36.3,51.1) | 44.0 (36.1,52.3) | 47.5 (14.0,83.4) | 100 (0,100) | 34.6 (19.1,54.3) | 0.931 |
| INS (%) | 37.0 (29.4,45.3) | 29.3 (23.1,36.3) | 34.3 (25.8,44.1) | 26.7 (10.4,53.3) | 0 (0,100) | 59.1 (22.8,87.6) | 0.568 |

Abbreviations: BMI, body mass index; HbA1c, glycated hemoglobin; FPG, fasting plasma glucose; 2-hour PG, 2-hour post-load glucose; TG, triglyceride; TC, total cholesterol; HDL-c, high-density lipoprotein cholesterol; LDL-c, low-density lipoprotein cholesterol; OHA, oral hypoglycemic drugs; INS, insulin.

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