Gene panel sequencing of patients with monogenic diabetes brings to light genes typically associated with syndromic presentations

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Supplementary Table 1: Clinical characteristics of the whole cohort.

Characteristic	
N	1676
Sex, female/male	54% / 46%
Eurocaucasian	47% (1583)
≥3 affected generations	52% (1421)
Age at diagnosis, years	25 [15-34] (1635)
BMI at diagnosis, kg/m²	23.5 [20.5-27.2] (1364)
BMI at diagnosis <25 kg/m²	848/514 (62%) (1362)
BMI at referral, kg/m²	24.2 [21.3-27.98] (1476)
Symptoms* at diagnosis	50% (1539)
HbA_{1c} at diagnosis, % HbA_{1c} at diagnosis, mmol/mol HbA_{1c} at referral, % HbA_{1c} at referral, mmol/mol	9.3 [6.8-12] (965) 78.15 [50.82-107.7] (965) 7.2 [6.3-8.9] (1355) 55.2 [45.36-73.78] (1355)
Duration since diagnosis, years	5 [1-15] (1636)
Therapy at diagnosis insulin/OHA/diet	593/546/355
Insulin therapy (at diagnosis): yes/no	40% (1494)
Therapy at referral insulin/OHA/diet	892/438/238
Insulin therapy (at referral): yes/no	57% (1568)
Arterial hypertension, yes/no	19% (1342)
Dyslipidemia, yes/no	27% (972)

*Symptoms defined by polyuria, weight loss, ketosis or ketoacidosis

Data are presented as median [25th percentile - 75th percentile] unless otherwise indicated. (n) total number of patients with available data.

Supplementary Table 2: Panel gene list.

Gene	OMIM ID	Reference transcript	Syndromic yes/no
ABCC8	600509	NM_001287174.1	no
CISD2	611507	NM_001008388.4	yes
GATA4	600576	NM_001308093.1	yes
GATA6	601656	NM_005257.5	yes
GCK	138079	NM_000162.3	no
HNF1A	142410	NM_000545.6	no
HNF1B	189907	NM_000458.3	yes
HNF4A	600281	NM_175914.4	no
INS	176730	NM_000207.2	no
INSR	147670	NM_000208.2	yes
KCNJ11	600937	NM_000525.3	no
NEUROD1	601724	NM_002500.4	no
PDX1	600733	NM_000209.4	no
PLIN1	170290	NM_002666.5	yes
RFX6	612659	NM_173560.3	no
TRMT10A	616013	NM_152292.4	yes
WFS1	606201	NM_006005.3	yes

Supplementary Table 3: List of variants identified in this study

***GCK* NM_000162.3 (104 unique heterozygous variants in 124 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.(?_1)_(45+1_46-1)del	p.?	CL5	1
c.(45+1_46-1)_(1398_?)del	p.?	CL5	1
c.(679+1_680-1)_(863+1_864-1)del	p.?	CL4	1
c.56T>A	p.Ile19Asn	CL4	1
c.106C>T	p.Arg36Trp	CL5	2
c.109A>G	p.Met37Val	CL4	1
c.110T>C	p.Met37Thr	CL4	1
c.113A>C	p.Gln38Pro	CL4	1
c.127C>T	p.Arg43Cys	CL5	1
c.128G>C	p.Arg43Pro	CL4	1
c.128G>A	p.Arg43His	CL5	1
c.130G>A	p.Gly44Ser	CL5	1
c.161G>A	p.Ser54Asn	CL4	1
c.184G>A	p.Val62Met	CL5	2
c.208+15C>G	p.?	CL3	1
c.214G>A	p.Gly72Arg	CL5	1
c.245C>T	p.Thr82Ile	CL4	1
c.266_275del	p.Val89fs	CL5	1
c.292C>T	p.Gln98*	CL5	2
c.329T>C	p.Ile110Thr	CL3	1
c.343A>G	p.Met115Val	CL4	1
c.364-1G>A	p.?	CL5	1
c.370G>C	p.Asp124His	CL4	1
c.386G>A	p.Cys129Tyr	CL5	1
c.446C>T	p.Thr149Ile	CL4	1
c.449T>C	p.Phe150Ser	CL5	1
c.452_454del	p.Ser151del	CL4	1

c.469G>A	p.Glu157Lys	CL1	2
c.478G>A	p.Asp160Asn	CL5	1
c.491T>A	p.Leu164His	CL4	1
c.508G>A	p.Gly170Ser	CL4	1
c.512T>G	p.Phe171Cys	CL4	1
c.526G>A	p.Ala176Thr	CL4	1
c.533G>A	p.Gly178Glu	CL4	1
c.544G>A	p.Val182Met	CL5	1
c.556C>T	p.Arg186*	CL5	1
c.559G>T	p.Asp187Tyr	CL4	1
c.562G>A	p.Ala188Thr	CL5	1
c.571C>T	p.Arg191Trp	CL5	6
c.572G>A	p.Arg191Gln	CL5	2
c.604A>G	p.Met202Val	CL3	1
c.607del	p.Val203*	CL4	1
c.607_627dup	p.Val203_Thr209dup	CL4	1
c.623C>T	p.Ala208Val	CL5	1
c.626C>A	p.Thr209Lys	CL4	1
c.629T>C	p.Met210Thr	CL5	2
c.635C>G	p.Ser212Cys	CL4	1
c.661G>A	p.Glu221Lys	CL5	2
c.667G>A	p.Gly223Ser	CL5	1
c.676G>A	p.Val226Met	CL4	2
c.679G>A	p.Gly227Ser	CL5	2
c.679+1G>T	p.?	CL5	1
c.679+5G>A	p.?	CL3	1
c.683C>A	p.Thr228Lys	CL4	1
c.683C>T	p.Thr228Met	CL5	1
c.700T>C	p.Tyr234His	CL4	1
c.713del	p.Met238fs	CL5	1
c.725A>G	p.Glu242Gly	CL4	1

c.748C>T	p.Arg250Cys	CL4	1
c.752T>C	p.Met251Thr	CL4	1
c.764C>G	p.Thr255Ser	CL4	1
c.764C>A	p.Thr255Asn	CL4	1
c.769T>C	p.Trp257Arg	CL5	2
c.773G>A	p.Gly258Asp	CL4	1
c.779T>G †	p.Phe260Cys	CL4	1
c.781G>A	p.Gly261Arg	CL5	2
c.782G>A	p.Gly261Glu	CL5	1
c.793G>A	p.Glu265Lys	CL5	1
c.823C>G	p.Arg275Gly	CL4	1
c.860A>T	p.Gln287Leu	CL5	1
c.895G>C	p.Gly299Arg	CL5	1
c.904G>A	p.Val302Met	CL4	1
c.908G>C	p.Arg303Pro	CL4	1
c.938dup	p.Asn313fs	CL5	1
c.952G>A	p.Gly318Arg	CL5	1
c.1007C>G	p.Ser336Trp	CL4	1
c.1019+1G>T	p.?	CL5	1
c.1020-1G>A	p.?	CL4	1
c.1025_1034delins	p.Thr342fs	CL5	1
c.1067G>T	p.Gly356Val	CL4	1
c.1068_1103del	p.Leu357_Arg368del	CL4	1
c.1072C>T	p.Arg358*	CL5	1
c.1112G>T	p.Cys371Phe	CL5	1
c.1118G>C	p.Ser373Thr	CL3	1
c.1118G>A	p.Ser373Asn	CL4	1
c.1121T>C	p.Val374Ala	CL4	1
c.1133C>A	p.Ala378Asp	CL4	1
c.1135G>A	p.Ala379Thr	CL4	1
c.1144T>G	p.Cys382Gly	CL5	1

c.1148C>T	p.Ser383Leu	CL5	1
c.1157_1160dup	p.Val389fs	CL5	1
c.1174C>T	p.Arg392Cys	CL4	1
c.1190G>A	p.Arg397His	CL4	1
c.1195G>T	p.Glu399*	CL5	2
c.1228G>A	p.Gly410Ser	CL4	1
c.1240A>G	p.Lys414Glu	CL5	1
c.1247A>C	p.His416Pro	CL4	1
c.1268T>A	p.Phe423Tyr	CL4	1
c.1318G>T	p.Glu440*	CL4	1
c.1322C>T	p.Ser441Leu	CL4	1
c.1324G>A	p.Glu442Lys	CL4	1
c.1340G>A	p.Arg447Gln	CL5	3
c.1348G>A	p.Ala450Thr	CL5	2
c.1358C>T	p.Ser453Leu	CL5	1
***HNF1A* NM_000545.6 (49 unique heterozygous variants in 63 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.(?-1)_(326+1_327-1)del	p.?	CL5	1
c.25C>T	p.Gln9*	CL5	1
c.98C>T	p.Pro33Leu	CL4	2
c.130del	p.Leu44fs	CL5	1
c.320T>C	p.Leu107Pro	CL5	1
c.388C>T	p.Gln130*	CL5	1
c.391C>T	p.Arg131Trp	CL5	2
c.392G>A	p.Arg131Gln	CL5	1
c.394G>A	p.Glu132Lys	CL4	1
c.457C>T	p.Pro153Ser	CL3	1
c.474G>T	p.Lys158Asn	CL5	1
c.475C>T	p.Arg159Trp	CL5	1
c.476G>A	p.Arg159Gln	CL5	5
c.498C>A	p.Tyr166*	CL4	1

c.509A>C	p.Gln170Pro	CL4	1
c.511C>G	p.Arg171Gly	CL4	1
c.517_526+5del	p.Val173fs	CL4	1
c.526C>T	p.Gln176*	CL5	1
c.526+1G>A	p.?	CL5	1
c.526+1G>C	p.?	CL5	1
c.527-1G>C	p.?	CL5	1
c.598C>T	p.Arg200Trp	CL5	1
c.599G>A	p.Arg200Gln	CL5	1
c.626C>A	p.Ala209Glu	CL3	1
c.685C>T	p.Arg229*	CL5	2
c.685C>G	p.Arg229Gly	CL4	1
c.690_691del	p.Glu230fs	CL5	1
c.754C>T	p.Gln252*	CL5	1
c.775G>A	p.Val259Ile	CL4	1
c.779C>T	p.Thr260Met	CL5	2
c.800G>A	p.Trp267*	CL4	1
c.811C>T	p.Arg271Trp	CL5	1
c.811C>G	p.Arg271Gly	CL4	1
c.812G>A	p.Arg271Gln	CL5	1
c.814C>A	p.Arg272Ser	CL5	1
c.815G>A	p.Arg272His	CL4	1
c.833G>A	p.Arg278Gln	CL4	1
c.872dupC	p.Gly292fs	CL5	6
c.966T>G	p.Tyr322*	CL5	1
c.1016G>A	p.Gly339Asp	CL3	1
c.1135C>A	p.Pro379Thr	CL4	1
c.1136delC	p.Pro379fs	CL5	1
c.1136_1137del	p.Pro379fs	CL5	2
c.1295C>T ‡	p.Ser432Phe	CL3	1
c.1333_1334del	p.Ser445fs	CL5	1

c.1340C>T	p.Pro447Leu	CL5	1
c.1475C>T	p.Thr492Ile	CL4	1
c.1502-1G>T	p.?	CL5	1
c.1740_1743dup	p.His582fs	CL4	1
***HNF4A* NM_175914.4 (27 unique heterozygous variants in 35 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.(?_-1)_(1359_?)del	p.?	CL5	1
c.(826+1_827-1)_(1063+1_1064-1)del	p.?	CL4	1
c.-181G>T	p.?	CL3	1
c.-173_-172del	p.?	CL3	1
c.124G>A	p.Gly42Arg	CL3	1
c.244del	p.Val82fs	CL5	1
c.322G>A	p.Val108Ile	CL5	1
c.334C>T	p.Arg112Trp	CL4	1
c.340C>T	p.Arg114Trp	CL5	5
c.352C>T	p.Arg118*	CL5	1
c.421C>T	p.Arg141*	CL5	2
c.502T>C	p.Ser168Pro	CL3	1
c.537G>A	p.Trp179*	CL5	1
c.572delT	p.Leu191fs	CL5	1
c.582+1G>C	p.?	CL5	1
c.583-2A>G	p.?	CL5	1
c.610G>A	p.Glu204Lys	CL3	1
c.617T>C	p.Leu206Pro	CL3	1
c.625G>A †	p.Gly209Arg	CL4	1
c.630_636del	p.Thr211fs	CL4	1
c.734G>A	p.Arg245His	CL4	2
c.869G>A	p.Arg290His	CL4	1
c.918T>G	p.Tyr306*	CL5	1
c.931C>T	p.Arg311Cys	CL5	3
c.949C>G	p.Leu317Val	CL4	1

c.977C>T	p.Thr326Ile	CL5	1
c.1063+1G>A	p.?	CL4	1
MT-TL1 **(1 ~~unique~~ heteroplasmic variant in 24 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
m.3243A>G	N/A	CL5	25
HNF1B **NM_000458.3 (8 unique heterozygous variants in 18 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.(?_-1)_(*1_?)del	p.?	CL5	12
c.344+2T>C	p.?	CL5	1
c.344+3A>G	p.?	CL3	1
c.409A>G	p.Arg137Gly	CL3	1
c.458A>G	p.His153Arg	CL5	1
c.476C>T	p.Pro159Leu	CL4	1
c.703C>T	p.Arg235Trp	CL4	1
ABCC8 **NM_001287174.1 (8 unique heterozygous variants in 8 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.395T>C	p.Phe132Ser	CL3	1
c.1220T>C	p.Leu407Pro	CL3	1
c.2588A>G	p.His863Arg	CL3	1
c.3527A>G	p.Tyr1176Cys	CL4	1
c.4135G>C ‡	p.Gly1379Arg	CL4	1
c.4139G>A	p.Arg1380His	CL5	1
c.4274T>C	p.Ile1425Thr	CL4	1
c.4435G>A	p.Gly1479Arg	CL4	1
PDX1 **NM_000209.4 (7 unique heterozygous variants in 7 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.98dup	p.Ala34fs	CL4	1
c.217dup	p.Leu73fs	CL4	1
c.532G>A	p.Glu178Lys	CL4	1
c.534_539del	p.Glu178_Ala180delinsAsp	CL3	1
c.559G>A	p.Glu187Lys	CL3	1

c.466_472dup	p.Leu158fs	CL3	1
c.697delG	p.Glu233fs	CL4	1
***RFX6* NM_173560.3 (7 unique heterozygous variants in 7 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.353del	p.Lys118fs	CL4	1
c.575_585del	p.Tyr192fs	CL5	1
c.872del	p.Leu291fs	CL4	1
c.1068G>A	p.Trp356*	CL4	1
c.1304_1307dup	p.Glu436fs	CL4	1
c.1513C>T	p.Arg505*	CL4	1
c.2499del	p.Tyr834fs	CL4	1
***NEUROD1* NM_002500.4 (5 unique heterozygous variants in 5 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.316G>A	p.Ala106Thr	CL4	1
c.321C>G	p.Asn107Lys	CL3	1
c.337C>T	p.Arg113Cys	CL3	1
c.340A>C	p.Met114Leu	CL4	1
c.616dupC	p.His206fs	CL5	1
***INS* NM_000207.2 (4 unique heterozygous variants in 4 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.16C>T	p.Arg6Cys	CL5	1
c.92G>T	p.Cys31Phe	CL5	1
c.100C>T	p.His34Tyr	CL4	1
c.130G>A	p.Gly44Arg	CL3	1
***INSR* NM_000208.2 (2 unique heterozygous variants in 2 probands)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.1548G>A	p.Trp516*	CL4	1
c.3602G>A	p.Arg1201Gln	CL4	1
***KCNJ11* NM_000525.3 (1 unique heterozygous variant in 1 proband)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.761C>A	p.Pro254Gln	CL4	1

***PLIN1* NM_002666.5 (1 unique heterozygous variant in 1 proband)**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.1191_1192del	p.Val398fs	CL4	1
***WFS1* NM_006005.3 (13 unique variants, either compound heterozygous or homozygous in 9 probands) §**			
DNA change	**Protein change**	**Classification**	**Number of probands**
c.124C>T	p.Arg42*	CL4	1
c.406C>T	p.Gln136*	CL4	1
c.670C>T	p.Gln224*	CL4	1
c.958_972del	p.Pro320_Ile324del	CL3	1
c.1037C>T	p.Pro346Leu	CL3	1
c.1220del	p.His407fs	CL4	1
c.1255_1260dup	p.Phe419_Pro420dup	CL3	1
c.1511C>G	p.Pro504Arg	CL4	1
c.1558C>T	p.Gln520*	CL5	1
c.1620_1622del	p.Trp540del	CL4	1
c.1672C>T	p.Arg558Cys	CL4	1
c.2104G>A	p.Gly702Ser	CL4	1
c.2207G>A	p.Gly736Asp	CL4	1

† one patient carried both the *GCK* c.779T>G p.Phe260Cys variant and the *HNF4A* c.625G>A p.Gly209Arg variant

‡ one patient carried both the *HNF1A* c.1295C>T p.Ser432Phe variant and the *ABCC8* c.4135G>C p.Gly1379Arg variant

§ Homozygous variants are indicated in grey. For genotypes of patients with compound heterozygous variants, see Table 2

Supplementary Table 4: Main characteristics of *GCK* patients at diabetes diagnosis

MgD patients with variants in :	*GCK*
N	124
Sex: female / men	65% / 35%
Eurocaucasian	83% (108)
≥3 affected generations	65% (114)
Age at diagnosis	15 [10-22] (119)
BMI at diagnosis, kg/m²	20.25 [16.4-22.7] (92)
BMI at referral, kg/m²	20.95 [18.55-24.4] (96)
Symptoms at diagnosis †	18% (116)
HbA_{1c} at diagnosis, % HbA_{1c} at diagnosis, mmol/mol HbA_{1c} at referral, % HbA_{1c} at referral, mmol/mol	6.3 [6-6.5] (84) 45.36 [42.35-47.55] (84) 6.25 [6-6.6] (90) 44.81 [41.81-59.57] (90)
Duration since diagnosis, years	1 [0-42] (119)
Therapy at diagnosis: insulin/OHA/diet	8/11/86
Insulin therapy (at diagnosis)	4% (101)
Therapy at referral insulin/OHA/diet	9/19/86
Insulin therapy (at referral)	8% (114)
Arterial hypertension	0% (98)
Dyslipidemia	3% (71)

Supplementary Table 5: Comparison of the clinical characteristics of patients with m.3243A>G mutation identified by Gene Panel Sequencing or by targeted m.3243A>G analysis

	Gene Panel Sequencing		**Targeted analysis of m.3243A>G**		
	Patients referred for monogenic diabetes		Patients referred for MIDD	Patients referred for monogenic diabetes*	
N	25		74	15	
Sex: female/male	48% / 52%		50% / 50%	60% / 40%	
Eurocaucasian	64% (25)		78% (54)	86% (14)	
Reported history of diabetes in the maternal family †	72% (25)		87% (55)	71% (14)	
Age at diagnosis, years	31 [27.5-38] (25)		35.5 [29.25-43] (68)	34 [24-39] (15)	
Symptoms of diabetes ‡	62.5% (24)		46% (57)	43% (14)	
BMI at diagnosis, kg/m²	21.08 [18.29-22.2] (22)		20.8 [18.6-22.7] (57)	20,57 [19.32-21.94] (12)	
Normal	91%		86%	100%	
overweight	9%		14%	-	
HbA_{1c} at diagnosis, %	10.25 [7.75-12.15] (16)		9.2 [7.5-11.9] (37)	7.1 [7-12.1] (11)	
HbA_{1c} at diagnosis, mmol/mol	88.53 [61.21-109.3] (16)		77.06 [58.48-106.6] (37)	54.1 [53.01-108.8] (11)	
Insulin therapy (at diagnosis)	41% (22)		34% (55)	13% (15)	
HbA_{1c} at referral, %	7.15 [6.1-8.375] (22)		7.5 [6.675-8.6] (54)	7.7 [6.658-8.65] (14)	
HbA_{1c} at referral, mmol/mol	54.65 [43.17-68.04] (22)		57.48 [49.46-70.5] (54)	60.66 [49.27-71.04] (14)	
Insulin therapy (at referral)	64% (25)		65% (63)	67% (15)	
	At referral	Post-diagnosis		At referral	Post-diagnosis
Deafness yes/not reported or yes/no/unevaluated *via* hearing test	5/20	14/1/10	64/1	0/15	3/0/9
Cardiomyopathy	1	4	11	1	1
Diabetes in the mother: yes/no/unevaluated	14/6/5		41/2/33	8/3/4	
Deafness in the mother: yes/unevaluated *via* hearing test	6/19		37/37	2/11	

* Replication cohort: 536 probands previously screened with a panel approach not including the m.3243A>G mutation, with age at diagnosis between 15 and 40 years old, BMI at diagnosis <25 (or when BMI at diagnosis was not available, BMI at referral < 25)

† Counting patients as long as a relative is diabetic in the maternal family (maternal grand-mother, maternal uncle…) even if the mother is not known to be diabetic

‡ Symptoms defined by polyuria, weight loss, ketosis or ketoacidosis

Data are presented as median [25th percentile - 75th percentile] unless otherwise indicated. (n) total number of patients with available data.

No parameter is significantly different between patients identified through Gene Panel Sequencing and those referred for MIDD

Supplementary Table 6: Comparison of the clinical characteristics of patients with an *HNF1B* mutation identified by Gene Panel Sequencing or by targeted analysis of the *HNF1B* gene

	Gene Panel Sequencing		Targeted analysis of *HNF1B*	p
N	18		145*	
Genotype: gene deletion / mutation	67% / 33%		60% / 40%	ns
Sex: female / male	44.4% / 55.6%		54.5% / 45.5%	ns
Eurocaucasian	82.4% (17)		83% (141)	ns
Parent with diabetes	93.3% (15)		52.1% (144)	< 0.0001
Reported familial history of renal disease†	16.7% (18)		33.8% (145)	ns
Age at diagnosis, years	27 [14.5-33.5] (18)		26 [18-34.25] (130)	ns
Symptoms of diabetes ‡	58.8% (17)		46.3% (121)	ns
BMI at diagnosis, kg/m²	22.9 [20.2-27.2] (15)		22.2 [19-24.1] (103)	ns
HbA_{1c} at diagnosis, % HbA_{1c} at diagnosis, mmol/mol	9.7 [6.85-13.65] (14) 82.52 [51.37-125.7] (14)		10.1 [6.7-14.7] (67) 87 [50-137] (67)	ns
Insulin therapy (at diagnosis)	61.1% (18)		49.6% (115)	ns
HbA_{1c} at referral, % HbA_{1c} at referral, mmol/mol	7.35 [6.52-9.05] (16) 56.84 [47.82-75.42] (16)		7.1 [6.2-8.1] (119) 54.1 [44.27-65.03] (119)	0.0244
Insulin therapy (at referral)	75% (16)		82.6% (115)	ns
Decreased renal function (at referral) §	5 (33.3%) (15)		35 (50.7%) (69)	ns
	At referral	Post-diagnosis		
Kidney morphological abnormalities	6	9	116 (127)	< 0.0001
Genital tract abnormalities	1	3	39 (61)	0.0005
Pancreas morphological abnormalities	2	4	50 (77)	0.0013
Intellectual/Learning disability	2	2	21 (93)	ns

Data are presented as median [25th percentile - 75th percentile] unless otherwise indicated. (n) total number of patients with available data. ns: not significant.

* reported in (1)

† kidney cysts or kidney failure

‡ Symptoms defined by polyuria, weight loss, ketosis or ketoacidosis

§ Chronic Kidney Disease stages 3 and 4, defined as an eGFR between 59 and 15 ml/min/1.73 m^2, or ESRD, end-stage renal disease, defined as an eGFR < 15ml/min/1.73 m^2, or dialysis or kidney transplantation.

1 - Dubois-Laforgue D, Cornu E, Saint-Martin C, Coste J, Bellanné-Chantelot C, Timsit J. Diabetes, Associated Clinical Spectrum, Long-term Prognosis, and Genotype/Phenotype Correlations in 201 Adult Patients With Hepatocyte Nuclear Factor 1B (*HNF1B*) Molecular Defects. Diabetes Care. 2017 Nov;40(11):1436–43.