

Gene OMIM # Cytogenetic location Genomic coordinates	Protein/ <u>Mode of inheritance</u>	MODY	Neonatal Diabetes Mellitus (NDM)	TNDM	PNDM	NDM autoimmunity	Syndromic	Reference
ABCC8 600509 11p15.1 11:17,392,884- 17,476,869	-ATP-binding cassette transporter sub-family C member 8 <u>-Spontaneous, dominant or recessive</u>	12	X		X		familial hyperinsulinemic hypoglycemia- I (HHF1)	<i>N Engl J Med</i> 355: 456-466, 2006. <i>Diabetologia</i> . 55(1):12 3-7, 2012.
APPL1 604299 3p14.3 3:57,227,736- 57,273,470	-Adaptor Protein, Phosphotyrosine Interaction, PH domain, and leucine zipper containing 1 <u>-Dominant</u>	14						<i>Am J Hum Genet</i> . 97(1):177-85, 2015.
BLK 191305 8p23.1 8:11,493,990- 11,564,598	-B lymphocyte kinase <u>-Dominant</u>	11						<i>PNAS</i> 106(34):14460- 5, 2009. <i>Diabetologia</i> . 56(3):49 2-6, 2013. <i>PLOS ONE</i> 12(1):1-15, 2017
BSCL2 606158 11q12.3 11:62,690,261- 62,709,618	-Seipin <u>-Recessive</u>		X				Congenital Generalized Lipodystrophy Type 2	<i>Nat Genet</i> . 28(4):365- 70, 2001.
CEL 114840 9q34.13 9:133,061,977- 133,071,862	-Carboxyl-ester lipase <u>-Dominant</u>	8						<i>Nat Genet</i> . 38(1):54- 62, 2006. <i>Hum Genet</i> . 127(1):55- 64, 2010.
DNAJC3 601184 13q32.1 13:95,677,138- 95,794,988	-Inhibitor of Protein Kinase, Interferon- Inducible Double-Stranded RNA-Dependent Kinase (PKR) PRKRI <u>-Recessive</u>						Diabetes Mellitus and Multisystemic Neurodegeneration	<i>Diabetes</i> 54: 1074- 1081, 2005. <i>Am. J. Hum. Genet</i> . 95: 689-697, 2014.
EIF2AK3 604032 2p11.2 2:88,556,739- 88,627,575	-Eukaryotic translation initiation factor 2-alpha kinase 3 (PERK) <u>-Recessive</u>		X		X		Wolcott-Rallison syndrome	<i>Nat Genet</i> . 25(4):406- 9, 2000.
FOXP3 300292 Xp11.23 X:49,250,435- 49,266,504	-forkhead box P3 <u>-X-linked, recessive</u>		X		X	X	Immune dysregulation, IPEX Syndrome, X- linked- IDDMX locus	<i>Nat Genet</i> . 27(1):18- 20, 2001.
GATA4 600576 8p23.1 8:11,676,918- 11,760,001	-Transcription factor GATA-4 <u>-Dominant</u>		X		X		Permanent neonatal diabetes with pancreatic agenesis and congenital heart defects	<i>Diabet Med</i> . 27(10):1195-200, 2010.
GATA6 601656 18q11.2 18:22,169,436- 22,202,527	-GATA-binding factor 6 <u>-Dominant</u>		X		X		pancreatic agenesis insulin-treated neonatal diabetes and exocrine pancreatic insufficiency	<i>Nat Genet</i> . 44(1):20-2, 2011. <i>Diabetes</i> . 62(3):993-7, 2013.
GCK 138079 7p13 7:44,143,212- 44,189,438	-Glucokinase <u>-Recessive</u>	2			X			<i>Nature</i> . 23:356(6371): 721-2, 1992.
GLIS3 610192 9p24.2 9:3,824,126- 4,310,693*	-GLIS Family Zinc Finger 3 <u>-Recessive</u>		X		X		Diabetes mellitus, neonatal, with congenital hypothyroidism; NDH syndrome	<i>Nat Genet</i> . 38(6):682- 7, 2006.
HNF1A 142410 12q24.31	-Hepatocyte nuclear factor 1-alpha <u>-Dominant</u>	3						<i>Nature</i> . 384(6608):455 -8, 1996. <i>Diabetes</i> . 46(4):720-5, 1997.

12:120,978,514-121,002,511								
HNF1B 189907 17q12 17:37,686,430-37,745,077	-Hepatocyte nuclear factor 1-beta <u>-Dominant</u>	5	X				Renal cysts and diabetes syndrome pancreatic agenesis	Nat Genet. 17(4):384-5, 1997.
HNF4A 600281 20q13.12 20:44,355,800-44,432,844	-Hepatocyte nuclear factor 4-alpha <u>-Dominant</u>	1						Nature. 384(6608):458-60, 1996.
IER3IP1 609382 18q21.1 18:47,155,018-47,176,373	-Immediate early response 3-interacting protein 1 <u>-Recessive</u>		X		X		Microcephaly, epilepsy, and diabetes syndrome; MEDS	Am J Hum Genet. 89(2):265-76, 2011.
IL2RA 147730 10p15.1 10:6,010,693-6,062,369**	-Interleukin-2 receptor alpha chain <u>-Susceptibility locus</u>		X			X	Immunodeficiency 41 lymphoproliferation and autoimmunity; IMD41	J Allergy Clin Immunol 119: 482-487, 2007
INS 176730 11p15.5 11:2,159,778-2,161,208***	-Insulin <u>-Spontaneous or dominant</u>	10	X	X	X			Diabetes. 57(4):1034-42, 2008
ITCH 606409 20q11.22 20:34,363,234-34,511,772	-E3 Ubiquitin Protein Ligase <u>-Recessive</u>					X	Autoimmune disease, multisystem, with facial dysmorphism; ADMFD	Am J Hum Genet. 86(3): 447-453, 2010.
KCNJ11 600937 11p15.1 11:17,385,245-17,389,330	-inward-rectifying ATP-sensitive K ⁺ channel <u>-Spontaneous or dominant</u>	13	X	X	X		Hyperinsulinemic Hypoglycemia	PLoS One. ;7(6): e37423, 2012
KLF11 603301 2p25.1 2:10,043,554-10,054,835	-Krueppel-like factor 11 <u>-Dominant</u>	7						J. Biol. Chem. 286, 28414-28424, 2011.
LRBA 606453 4q31.3 4:150,264,514-151,015,724	-Lipopolysaccharide-responsive and beige-like anchor protein <u>-Recessive</u>		X			X	common variable immunodeficiency-8 with autoimmunity; CVID8	J Clin Endocrinol Metab. 101(3):898-904, 2016.
MX1 142994 7q36.3 7:157,004,852-157,010,652	-motor neuron and pancreas homeobox 1 <u>-Recessive</u>		X		X		Neonatal Diabetes; Currarino Syndrome	Diabetes Metab. 39(3):276-80, 2013. Cell Metab. 19(1):146-54, 2014.
NEUROD1 601724 2q31.3 2:181,676,105-181,680,664	-Neurogenic differentiation 1 <u>-Recessive</u>	6	X		X			Diabetes 59(9):2326-31, 2010.
NEUROG3 604882 10q22.1 10:69,571,439-69,573,453	-Neurogenin-3 <u>-Recessive</u>		X		X		childhood-onset diabetes; Permanent Neonatal Diabetes and Enteric Anendocrinosis; DIAR4	Diabetes 60(4):1349-1353, 2011.
NKX2-2 604612 20p11.22 20:21,511,009-21,514,025	-Homeobox protein Nkx-2.2 <u>-Recessive</u>		X		X		Neonatal Diabetes	Cell Metabolism 19:146-154, 2014.
NR0B2 604630 1p36.11	-Nuclear Receptor Subfamily 0 Group B Member 2 (SHP)		X				Diabetes associated with mild to moderate obesity	PNAS 98:575-580, 2001.

1:26,911,483-26,914,075	-Dominant (obesity not diabetes)							
PAX4 167413 7q32.1 7:127,610,291-127,618,191	-Paired box gene 4 -Dominant	9						Acta Diabetol. 53(2):205-16, 2016.
PAX6 607108 11p13 11:31,784,791-31,817,960	-Paired box protein Pax-6 -Recessive						Aniridia and Glucose Intolerance	Diabetes 51:224-230, 2002.
PCBD1 126090 10q22.1 10:70,882,279-70,888,785	-pterin-4 α -carbinolamine dehydratase/dimerization cofactor of hepatocyte nuclear factor 1 α -Recessive		X		X		Early-onset non-autoimmune diabetes with features similar to dominantly inherited HNF1A-diabetes	Diabetes 63(10):3557-64, 2014.
PDX1 600733 13q12.2 13:27,919,981-27,926,313	-Pancreatic duodenal homeobox 1 -Recessive	4	X		X		Permanent neonatal diabetes +/- pancreatic agenesis	Diabet Med. 30(5):e197-200, 2013.
PLAGL1 603044 6q24.2 6:143,940,299-144,064,598	-Pleomorphic adenoma gene-like 1 -Variable (imprinting)		X	X			transient neonatal diabetes mellitus.	Hum. Molec. Genet. 9:453-460, 2000. Hum. Genet. 110: 139-144, 2002.
PTF1A 607194 10p12.2 10:23,192,530-23,194,251	-Pancreas transcription factor 1 subunit alpha -Recessive		X		X		cerebellar and pancreatic agenesis	Nat Genet. 36(12):1301-5, 2004.
RFX6 612659 6q22.1 6:116,877,212-116,932,162	-Regulatory factor X, 6 -Recessive		X		X		Permanent neonatal diabetes; pancreatic hypoplasia, intestinal atresia, gallbladder aplasia or hypoplasia: Mitchell-Riley Syndrome	Nature. 463(7282):775-80, 2010.
SIRT1 604479 10q21.3 10:67,884,668-67,918,389	-NAD-dependent deacetylase sirtuin-1 --Dominant					X	Late onset 7,12, 15 and 27 yo T1D diagnosis	Cell Metabolism 17:448-455, 2013.
SLC19A2 603941 1q24.2 1:169,463,908-169,485,969	-solute carrier family 19 member 2 (Thiamine transporter 1) -Recessive		X		X		Thiamine responsive megaloblastic anaemia, diabetes and deafness (TRMA) syndrome	Pediatric Diabetes 13(4):314-21, 2012. Nat Genet 22: 300-304, 1999
SLC2A2 138160 3q26.2 3:170,996,340-171,026,978	-solute carrier family 2 member 2 (GLUT2) -Recessive		X	X			Fanconi-Bickel Syndrome	Nat Genet. 17(3):324-6, 1997.
STAT1 600555 2q32.2 2:190,969,035-191,014,249	-Signal transducer and activator of transcription 1 -Dominant					X	Immunodeficiency 31A,C; Gain-of-function mutations in STAT1 can cause an IPEX-like phenotype	J Allergy Clin Immunol. 131(6):1611-23, 2013
STAT3 102582 17q21.2 17:42,313,323-42,388,504	-Signal transducer and activator of transcription 3 -Dominant		X		X	X	Infantile-Onset Multisystem Autoimmune Disease 1 (ADMIO1)	Blood 125:591-59, 2015. Nat Genet. 46(8):812-4, 2014.
STAT5B 604260 17q21.2 17:42,199,176-42,276,459	-Signal transducer and activator of transcription 5					X	Growth hormone insensitivity with immunodeficiency	Ann N Y Acad Sci. 1079:198-204, 2006 Diabetes 55(10):2705-12, 2006

								<i>J Biol Chem.</i> 279:11553-61, 2004
TRMT10A 616013 4q23 4:99,546,706- 99,564,056	-tRNA methyltransferase homolog gene -Recessive		X				Microcephaly, short stature, and impaired glucose metabolism 1	<i>PLOS Genetics</i> 9:1-15, 2013.
WFS1 606201 4p16.1 4:6,260,367- 6,303,264	-Wolframin -Recessive						Wolfram syndrome (Diabetes insipidus, diabetes mellitus, optic atrophy and deafness, DIDMOAD)	<i>Nat Genet.</i> 20(2):143- 8, 1998.
ZFP57 612192 6p22.1 6:29,672,391- 29,681,149	-Zinc finger protein 57 homolog -Recessive		X	X			Transient neonatal diabetes	<i>Nat Genet.</i> 40(8):949- 5, 2008.

Table S3

The annotation of this table was derived from data associated with the University of Exeter Molecular Genetics Laboratory (http://www.diabetesgenes.org/sites/default/files/tngs_genes_v4.pdf) and the Online Mendelian Inheritance in Man[®] (OMIM[®]) site (<https://www.omim.org/>). Genomic coordinates are derived from the reference genome GRCh38.

* The GLIS3 gene associates with the markers: rs10758593; rs7020673; and rs6476839 (Immunobase <https://www.immunobase.org/disease/T1D/>).

** The IL2RA gene associates with the markers:

rs61839660; rs12251307; rs2104286; rs41295121; rs7090530; and rs10795791.

*** The INS gene associates with the markers:

rs72853903; rs689; rs7111341; and rs7928968.