Royal Devon and Exeter NHS Foundation Trust

UKAS MEDICAL 8092

Exeter Molecular Genetics Laboratory
Monogenic Diabetes Gene Panel v5.3

Gene (OMIM)	Genbank Reference Sequence	Phenotype	OMIM	Inheritance	References
ABCC8 600509	NM_001287174	Permanent neonatal diabetes	606176	Dominant (often *de novo*) or recessive	Proks *et al* 2006 Hum Mol Genet 15: 1793-1800 Babenko *et al* 2006 N Engl J Med 355: 456-466 Ellard *et al* 2007 Am J Hum Genet 81: 375-382
		Transient neonatal diabetes	610374	Dominant (often *de novo*) or recessive	Babenko *et al* 2006 N Engl J Med 355: 456-466
		MODY	610374	Dominant	Bowman *et al* 2012 Diabetologia 55: 123-127 Riveline *et al* 2012 Diabetes Care 35: 248-251
AGPAT2 603100	NM_006412	Congenital generalised lipodystrophy	608594	Recessive	Agarwal *et al* 2002 Nat Genet 31: 21-23
AIRE 607358	NM_000383	Type 1 autoimmune polyendocrinopathy syndrome and autoimmune diabetes	240300	Dominant (often *de novo*) or recessive	Finnish-German APECED Consortium 1997 Nat Genet 17: 399-403
AKT2 164731	NM_001626	Lipodystropy and severe insulin resistance	Not assigned	Dominant	George *et al* 2004 Science 304: 1325-1328 Semple *et al* 2009 J Clin Invest 119: 315-322
APPL1 604299	NM_012096	MODY	616511	Dominant	Prudente *et al* 2015 Am J Hum Genet 97: 177-185
BSCL2 606158	NM_032667	Congenital generalised lipodystrophy, severe insulin resistance and diabetes	269700	Recessive	Magre *et al* 2001 Nat Genet 28: 365-370
CTLA4 123890	NM_005214	Type V autoimmune lymphoproliferative syndrome and autoimmune diabetes	616100	Dominant	Schubert *et al* 2014 Nature Med 20: 1410-1416 Kuehn *et al* 2014 Science 345: 1623-1627

CEL 114840	NM_001807	MODY	609812	Dominant	Raeder *et al* 2006 Nat Genet 38: 54-62 Torsvik *et al* 2010 Hum Genet 127: 55-64 Raeder *et al* 2013 PLoS One 8: e60229
CISD2 611507	NM_001008388	Wolfram Syndrome 2 (diabetes mellitus, hearing loss, optic atrophy and defective platelet aggregation).	604928	Recessive	Amr *et al* 2007 Amr J Hum Genet 81: 673-683
COQ2 609825	NM_015697	Coenzyme Q10 deficiency, primary, 1 (hyperglycaemia reported)	607426	Recessive	Quinzii *et al* 2006 Am J Hum Genet 78: 345-349
COQ9 612837	NM_020312	Coenzyme Q10 deficiency, primary, 5 (hyperglycaemia reported)	614654	Recessive	Duncan *et al* 2009 Am J Hum Genet 84: 558-566
DCAF17 612515	NM_025000	Woodhouse-Sakati syndrome	241080	Recessive	Alazami *et al* 2008 Am J Hum Genet 83: 684-691
DNAJC3 601184	NM_006260	Autosomal recessive juvenile-onset diabetes with central and peripheral neurodegeneration	616192	Recessive	Synofzik *et al* 2014 Am J Hum Genet 95: 689-697
DYRK1B 604556	NM_004714	Diabetes and metabolic syndrome	615812	Dominant	Keramati *et al* 2014 New Eng J Med 370: 1909-1919
EIF2AK3 604032	NM_004836	Wolcott-Rallison syndrome	226980	Recessive	Delephine *et al* 2000 Nat Genet 25: 406-409
EIF2S3 300161	NM_001415	Borck type of X-linked syndromic mental retardation and neonatal diabetes	300987	X-Linked Recessive	Moortgat *et al* 2016 Am J Med Genet 170A: 2927-2933
FOXP3 300292	NM_014009	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked syndrome (IPEX)	304790	X-Linked Recessive	Wildin *et al* 2001 Nat Genet 1: 18-20 Bennett *et al* 2001 Nat Genet 27: 20-21
GATA4 600576	NM_002052	Permanent neonatal diabetes with pancreatic agenesis and congenital heart defects	Not assigned	Dominant (often *de novo*)	D'Amato *et al* 2010 Diabet Med 27: 1195-1200

GATA6 601656	NM_005257	Permanent neonatal diabetes with pancreatic agenesis and congenital heart defects	600001	Dominant (often *de novo*)	Lango Allen *et al* 2011 Nat Genet 44: 20-22 De Franco *et al* 2013 Diabetes 62: 993-997
GCK 138079	NM_000162	Permanent neonatal diabetes	606176	Recessive	Njolstad *et al* 2001 N Engl J Med 344: 1588-1592 Gloyn *et al* 2002 Diabetologia 45: 290 Osbak *et al* 2009 Hum Mutat 30: 1512-1526
		MODY	125851	Dominant	Vionnet *et al* 1992 Nature 356: 721-722 Velho *et al* 1997 Diabetologia 40: 217-224 Osbak *et al* 2009 Hum Mutat 30: 1512-1526
GLIS3 610192	NM_001042413	Permanent neonatal diabetes with congenital hypothyroidism	610199	Recessive	Senee *et al* 2006 Nat Genet 38: 682-687 Dimitri *et al* 2011 Eur J Endocrinol 164: 437-443
HNF1A 142410	NM_000545	MODY	600496	Dominant	Yamagata *et al* 1996 Nature 384: 455-458 Frayling *et al* 1997 Diabetes 46: 720-725 Colclough *et al* 2013 Hum Mutat 34: 669-685
HNF1B 189907	NM_000458	Renal Cysts and Diabetes syndrome (RCAD)	137920	Dominant (often *de novo*)	Horikawa *et al* 1997 Nat Genet 17: 384-385 Yorifuji *et al* 2004 J Clin Endocrinol Metab 89: 2905-2908 Edghill *et al* 2006 J Med Genet 43: 84-90 Bellanne-Chantelot *et al* 2005 Diabetes 54: 3126-3132
HNF4A 600281	NM_175914	MODY	125850	Dominant	Yamagata *et al* 1996 Nature 384: 458-460 Bulman *et al* 1997 Diabetologia 40: 859-862 Colclough *et al* 2013 Hum Mutat 34: 669-685
IER3IP1 609382	NM_016097	microcephaly, epilepsy, and diabetes syndrome (MEDS)	614231	Recessive	Poulton et al 2011 Am J Hum Genet 89: 265-276 Abdel-Salam *et al* 2012 Am J Med Genet A 158A: 2788-2796
IL2RA 147730	NM_000417	Immunodeficiency 41 with lymphoproliferation, autoimmunity and autoimmune diabetes	606367	Recessive	Caudy *et al* 2007 J Allergy Clin Immunol 119: 482-487
INS	NM_001185098	Permanent neonatal	606176	Dominant	Stoy *et al* 2007 Proc Natl Acad Sci USA 18: 15040-15044

176730		diabetes		(often *de novo*) or recessive	Edghill *et al* 2008 Diabetes 57: 1034-1042
		Transient neonatal diabetes	Not assigned	Dominant (often *de novo*) or recessive	Garin *et al* 2010 Proc Natl Acad Sci 107: 3105-3110
		MODY	613370	Dominant	Edghill *et al* 2008 Diabetes 57: 1034-1042 Molven *et al* 2008 Diabetes 57: 1131-1135
INSR 147670	NM_000208	Severe insulin resistance	610549	Dominant	Odawara *et al* 1989 Science 245: 66-68
ITCH 606409	NM_001257138	Multisystem autoimmune disease with facial dysmorphism and autoimmune diabetes	613385	Recessive	Lohr *et al* 2010 Am J Hum Genet 86: 447-453
JAK1 147795	NM_002227	Immune dysregulatory and hypereosinophilic syndrome	Not assigned	Dominant	Del Bel *et al* 2017 J Allergy Clin Immunol 139: 2016-2020
KCNJ11 600937	NM_000525	Permanent neonatal diabetes	606176	Dominant (often *de novo*)	Gloyn *et al* 2004 N Engl J Med 350: 1838-1849
		Transient neonatal diabetes	610582	Dominant (often *de novo*)	Yorifuji *et al* 2005 J Clin Endocrinol Metab 90: 3174-3178 Gloyn *et al* 2005 Hum Mol Genet 14: 925-934 Suzuki *et al* 2007 J Clin Endocrinol Metab 92: 3979-3985
		MODY	616329	Dominant	Yorifuji *et al* 2005 J Clin Endocrinol Metab 90: 3174-3178 Bonnefond *et al* 2012 PLoS One 7: e37423
LMNA 150330	NM_170707	Familial Partial Lipodystropy (FPLD2) and insulin resistance	151660	Dominant	Cao *et al* 2000 Hum Mol Genet 1: 109-112 Shackleton *et al* 2000 Nat Genet 24: 153-156 Speckman *et al* 2000 Am J Hum Genet 66: 1192-1198
LPL 609708	NM_00237	Lipoprotein lipase deficiency and transient neonatal diabetes	238600	Recessive	Raupp *et al* 2002 J Inherit Metab Dis 25: 413-414
LRBA 606453	NM_001199282	Immunodysregulation and autoimmune diabetes	606453	Recessive	Charbonnier *et al* 2015 J Allergy Clin Immunol 135: 217-227 Schreiner *et al* 2016 J Clin Endocrinol Metab 101: 898-904
MNX1 142994	NM_005515	Neonatal diabetes & IUGR	Not assigned	Recessive	Flanagan *et al* 2014 Cell Metab 19: 146-154

***MTTL1* g.3243A>G** 590050	NC_012920	Maternally inherited diabetes and deafness (MIDD)	520000	Mitochondrial	Van den Ouweland *et al* 1992 Nat Genet 1: 368-371 Murphy *et al* 2008 Diabet Med 25: 383-399
NEUROD1 601724	NM_002500	Permanent neonatal diabetes and neurological abnormalities	Not assigned	Recessive	Rubio-Cabezas *et al* 2010 Diabetes 162: 987-992
		MODY	606394	Dominant	Malecki *et al* 1999 Nat Genet 23: 323-328
NEUROG3 604882	NM_020999	Permanent neonatal diabetes with congenital malabsorptive diarrhoea	610370	Recessive	Rubio-Cabezas *et al* 2011 Diabetes 60: 1349-1353
NKX2-2 604612	NM_002509	Neonatal diabetes and developmental delay	Not assigned	Recessive	Flanagan *et al* 2014 Cell Metab 19: 146-154
PAX6 607108	NM_001604	Aniridia and impaired glucose tolerance	106210	Dominant	Yasuda *et al* 2002 Diabetes 51: 224-230 Nishi *et al* 2005 Diabet Med 22: 641-644 Osawa *et al* 2015 J Diabetes Investig 6: 105-106
PCBD1 126090	NM_000281	MODY	Not assigned	Recessive	Simaite *et al* 2014 Diabetes 63: 3557-3564 Ferre *et al* 2014 J Am Soc Nephrol 25: 574-586
PDX1 600733	NM_000209	Permanent neonatal diabetes +/- pancreatic agenesis	260370	Recessive	Stoffers *et al* 1997 Nat Genet 15: 106-110 Thomas *et al* 2009 Pediatr Diabetes 10: 492-496 De Franco *et al* 2013 Diabet Med 30: e197-200
		MODY	606392	Dominant	Stoffers *et al* 1997 Nat Genet 17: 138-139
PIK3R1 171833	NM_181523	SHORT syndrome	269880	Dominant	Dyment *et al* 2013 Am J Hum Genet 93: 158-166
PLIN1 170290	NM_002666	Familial Partial Lipodystropy (FPLD4) and insulin resistance	613877	Dominant	Gandotra *et al* 2011 N Engl J Med 364: 740-748
POLD1 174761	NM_002691	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy (MDPL) syndrome	615381	Dominant (*de novo*)	Weedon *et al* 2013 Mat Genet 45: 947-950
PPARG 601487	NM_015869	Familial Partial Lipodystropy (FPLD3) and insulin resistance	604367	Dominant	Agarwal *et al* 2002 J Clin Endocrinol Metab 1: 408-411 Barroso *et al* 1999 Nature 402: 880-883
PPP1R15B 613257	NM_032833	Juvenile-onset diabetes with microcephaly, epilepsy and	616817	Recessive	Abdulkarim *et al* 2015 Diabetes 64: 3951-3962

		intellectual disability			
PTF1A 607194	NM_178161	Permanent neonatal diabetes with cerebellar and pancreatic agenesis	609069	Recessive	Sellick *et al* 2004 Nat Genet 36: 1301-1305
RFX6 612659	NM_173560	Permanent neonatal diabetes with pancreatic hypoplasia, intestinal atresia, and gallbladder aplasia or hypoplasia	601346	Recessive	Smith *et al* 2010 Nature 463: 775-780
SIRT1 604479	NM_012238	Monogenic autoimmune autoimmune diabetes	Not assigned	Dominant	Biason-Lauber *et al* 2013 Cell Metab. 17: 448-455
SLC2A2 138160	NM_000340	Fanconi-Bickel syndrome	227810	Recessive	Santer *et al* 1997 Nat Genet 17: 324-326
SLC19A2 603941	NM_006996	Thiamine responsive megaloblastic anaemia, diabetes and deafness (TRMA) syndrome	249270	Recessive	Labay *et al* 1999 Nat Genet 22: 300-304
SLC29A3 612373	NM_018344	H syndrome & PHID syndrome	602782	Recessive	Cliffe *et al* 2009 Hum Molec Genet 18: 2257-2265 Molho-Pessach *et al* 2008 Am J Hum Genet 83: 529-534
STAT1 600555	NM_007315	Immunodeficiency 31C and IPEX-like phenotype	614162	Dominant	Uzel *et al* 2013 J Allergy Clin Immun 131: 1611-1623
STAT3 102582	NM_139276	Neonatal diabetes and polyautoimmune disease	615952	Dominant	Flanagan *et al* 2014 Nat Genet 46: 812-814
STAT5B 604260	NM_012448	Growth hormone insensitivity with immunodeficiency	245590	Recessive	Kofoed *et al* 2003 N Engl J Med 349: 1139-147
TNFAIP3 191163	NM_001270508	Familial Behcet-Like Autoinflammatory Syndrome and autoimmune diabetes	616744	Dominant	Zhou *et al* 2016 Nat Genet 48: 67-73
TRMT10A 616013	NM_001134665	Juvenile-onset diabetes with microcephaly, epilepsy and intellectual disability	616033	Recessive	Igoillo-Esteve *et al* 2013 PLoS Genet 9: e1003888
WFS1 606201	NM_006005	Wolfram syndrome (Diabetes insipidus, diabetes mellitus, optic atrophy and deafness, DIDMOAD)	222300	Recessive	Inoue *et al* 1998 Nat Genet 20: 143-148 Strom *et al* 1998 Hum Mol Genet 7: 2021-2028
ZBTB20 606025	NM_001164342	Primrose syndrome	259050	Dominant (*de novo*)	Cordeddu *et al* 2014 Nat Genet 46: 815-817

ZFP57 612192	NM_001109809	Transient neonatal diabetes	601410	Recessive	Mackay *et al* 2008 Nat Genet 40: 949-951