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Royal Devon and Exeter

NHS Foundation Trust



Exeter Molecular Genetics Laboratory

Monogenic Diabetes Gene Panel v5.4

Gene (OMIM)	Genbank Reference Sequence	Phenotype	OMIM	Inheritance	References
ABCC8 600509	NM_001287174	Permanent neonatal diabetes	606176	Dominant (often <i>de novo</i>) or recessive	Proks <i>et al</i> 2006 Hum Mol Genet <u>15</u> : 1793-1800 Babenko <i>et al</i> 2006 N Engl J Med <u>355</u> : 456-466 Ellard <i>et al</i> 2007 Am J Hum Genet <u>81</u> : 375-382
		Transient neonatal diabetes	610374	Dominant (often <i>de novo</i>) or recessive	Babenko <i>et al</i> 2006 N Engl J Med <u>355</u> : 456-466
		MODY	610374	Dominant	Bowman <i>et al</i> 2012 Diabetologia <u>55</u> : 123-127 Riveline <i>et al</i> 2012 Diabetes Care <u>35</u> : 248-251
AGPAT2 603100	NM_006412	Congenital generalised lipodystrophy	608594	Recessive	Agarwal <i>et al</i> 2002 Nat Genet <u>31</u> : 21-23
AIRE 607358	NM_000383	Type 1 autoimmune polyendocrinopathy syndrome and autoimmune diabetes	240300	Dominant (often <i>de novo</i>) or recessive	Finnish-German APECED Consortium 1997 Nat Genet <u>17</u> : 399-403
AKT2 164731	NM_001626	Lipodystrophy and severe insulin resistance	Not yet assigned	Dominant	George <i>et al</i> 2004 Science <u>304</u> : 1325-1328 Semple <i>et al</i> 2009 J Clin Invest <u>119</u> : 315-322
APPL1 604299	NM_012096	MODY	616511	Dominant	Prudente <i>et al</i> 2015 Am J Hum Genet <u>97</u> : 177-185
BSCL2 606158	NM_032667	Congenital generalised lipodystrophy, severe insulin resistance and diabetes	269700	Recessive	Magre <i>et al</i> 2001 Nat Genet <u>28</u> : 365-370
CD55 125240	NM_000574	Complement hyperactivation, angiopathic thrombosis and protein-losing enteropathy	226300	Recessive	Kurolap <i>et al</i> 2017 New Eng J Med <u>377</u> : 87-89 Ozen <i>et al</i> 2017 New Eng J Med <u>377</u> : 52-61

CEL 114840	NM_001807	MODY and pancreatic exocrine dysfunction	609812	Dominant	Raeder <i>et al</i> 2006 Nat Genet <u>38</u> : 54-62 Torsvik <i>et al</i> 2010 Hum Genet <u>127</u> : 55-64 Raeder <i>et al</i> 2013 PLoS One <u>8</u> : e60229
CIDEC 612120	NM_001199623	Familial Partial Lipodystrophy (FPLD5) and insulin resistance	615238	Recessive	Rubio-Cabezas <i>et al</i> 2009 EMBO Mol Med <u>1</u> : 280-287
CISD2 611507	NM_001008388	Wolfram Syndrome 2 (diabetes mellitus, hearing loss, optic atrophy and defective platelet aggregation).	604928	Recessive	Amr <i>et al</i> 2007 Amr J Hum Genet <u>81</u> : 673-683
CNOT1 604917	NM_016284	holoprosencephaly-12 with pancreatic agenesis and neonatal diabetes	618500	Dominant	De Franco <i>et al</i> 2019 Am J Hum Genet <u>104</u> : 985-989
COQ2 609825	NM_015697	Coenzyme Q10 deficiency, primary, 1 (hyperglycaemia reported)	607426	Recessive	Quinzii <i>et al</i> 2006 Am J Hum Genet <u>78</u> : 345-349
COQ9 612837	NM_020312	Coenzyme Q10 deficiency, primary, 5 (hyperglycaemia reported)	614654	Recessive	Duncan <i>et al</i> 2009 Am J Hum Genet <u>84</u> : 558-566
CTLA4 123890	NM_005214	Type V autoimmune lymphoproliferative syndrome and autoimmune diabetes	616100	Dominant	Schubert <i>et al</i> 2014 Nature Med <u>20</u> : 1410-1416 Kuehn <i>et al</i> 2014 Science <u>345</u> : 1623-1627
DCAF17 612515	NM_025000	Woodhouse-Sakati syndrome	241080	Recessive	Alazami <i>et al</i> 2008 Am J Hum Genet <u>83</u> : 684-691
DNAJC3 601184	NM_006260	Autosomal recessive juvenile-onset diabetes with central and peripheral neurodegeneration	616192	Recessive	Synofzik <i>et al</i> 2014 Am J Hum Genet <u>95</u> : 689-697
DOCK8 611432	NM_203447	Hyper-IgE recurrent infection syndrome & combined immunodeficiency	243700	Recessive	Zhang <i>et al</i> 2009 New Eng J Med <u>361</u> : 2046-2055
DUT 601266	NM_001025248	Diabetes and bone marrow failure	Not yet assigned	Recessive	Dos Santos <i>et al</i> 2017 Diabetes <u>66</u> : 1086-1096
DYRK1B 604556	NM_004714	Diabetes and metabolic syndrome	615812	Dominant	Keramati <i>et al</i> 2014 New Eng J Med <u>370</u> : 1909-1919

EIF2AK3 604032	NM_004836	Wolcott-Rallison syndrome	226980	Recessive	Delephine <i>et al</i> 2000 Nat Genet <u>25</u> : 406-409
EIF2S3 300161	NM_001415	Borck type of X-linked syndromic mental retardation and neonatal diabetes	300987	X-Linked Recessive	Moortgat <i>et al</i> 2016 Am J Med Genet <u>170A</u> : 2927-2933
FOXP3 300292	NM_014009	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked syndrome (IPEX)	304790	X-Linked Recessive	Wildin <i>et al</i> 2001 Nat Genet <u>1</u> : 18-20 Bennett <i>et al</i> 2001 Nat Genet <u>27</u> : 20-21
GATA4 600576	NM_002052	Permanent neonatal diabetes with pancreatic agenesis and congenital heart defects	Not yet assigned	Dominant (often <i>de novo</i>)	D'Amato <i>et al</i> 2010 Diabet Med <u>27</u> : 1195-1200
GATA6 601656	NM_005257	Permanent neonatal diabetes with pancreatic agenesis and congenital heart defects	600001	Dominant (often <i>de novo</i>)	Lango Allen <i>et al</i> 2011 Nat Genet <u>44</u> : 20-22 De Franco <i>et al</i> 2013 Diabetes <u>62</u> : 993-997
GCK 138079	NM_000162	Permanent neonatal diabetes	606176	Recessive	Njolstad <i>et al</i> 2001 N Engl J Med <u>344</u> : 1588-1592 Gloyn <i>et al</i> 2002 Diabetologia <u>45</u> : 290 Osbak <i>et al</i> 2009 Hum Mutat <u>30</u> : 1512-1526
		MODY	125851	Dominant	Vionnet <i>et al</i> 1992 Nature <u>356</u> : 721-722 Velho <i>et al</i> 1997 Diabetologia <u>40</u> : 217-224 Osbak <i>et al</i> 2009 Hum Mutat <u>30</u> : 1512-1526
GLIS3 610192	NM_001042413	Permanent neonatal diabetes with congenital hypothyroidism	610199	Recessive	Senee <i>et al</i> 2006 Nat Genet <u>38</u> : 682-687 Dimitri <i>et al</i> 2011 Eur J Endocrinol <u>164</u> : 437-443
HNF1A 142410	NM_000545	MODY	600496	Dominant	Yamagata <i>et al</i> 1996 Nature <u>384</u> : 455-458 Frayling <i>et al</i> 1997 Diabetes <u>46</u> : 720-725 Colclough <i>et al</i> 2013 Hum Mutat <u>34</u> : 669-685
HNF1B 189907	NM_000458	Renal Cysts and Diabetes syndrome (RCAD)	137920	Dominant (often <i>de novo</i>)	Horikawa <i>et al</i> 1997 Nat Genet <u>17</u> : 384-385 Yorifuji <i>et al</i> 2004 J Clin Endocrinol Metab <u>89</u> : 2905-2908 Edghill <i>et al</i> 2006 J Med Genet <u>43</u> : 84-90

					Bellanne-Chantelot <i>et al</i> 2005 Diabetes 54 : 3126-3132
HNF4A 600281	NM_175914	MODY	125850	Dominant	Yamagata <i>et al</i> 1996 Nature 384 : 458-460 Bulman <i>et al</i> 1997 Diabetologia 40 : 859-862 Colclough <i>et al</i> 2013 Hum Mutat 34 : 669-685
IER3IP1 609382	NM_016097	microcephaly, epilepsy, and diabetes syndrome (MEDS)	614231	Recessive	Poulton <i>et al</i> 2011 Am J Hum Genet 89 : 265-276 Abdel-Salam <i>et al</i> 2012 Am J Med Genet A 158A : 2788-2796
IL2RA 147730	NM_000417	Immunodeficiency 41 with lymphoproliferation, autoimmunity and autoimmune diabetes	606367	Recessive	Caudy <i>et al</i> 2007 J Allergy Clin Immunol 119 : 482-487
INS 176730	NM_001185098	Permanent neonatal diabetes	606176	Dominant (often <i>de novo</i>) or recessive	Stoy <i>et al</i> 2007 Proc Natl Acad Sci USA 18 : 15040-15044 Edghill <i>et al</i> 2008 Diabetes 57 : 1034-1042
		Transient neonatal diabetes	Not yet assigned	Dominant (often <i>de novo</i>) or recessive	Garin <i>et al</i> 2010 Proc Natl Acad Sci 107 : 3105-3110
		MODY	613370	Dominant	Edghill <i>et al</i> 2008 Diabetes 57 : 1034-1042 Molven <i>et al</i> 2008 Diabetes 57 : 1131-1135
INSR 147670	NM_000208	Severe insulin resistance	610549	Dominant	Odawara <i>et al</i> 1989 Science 245 : 66-68
ITCH 606409	NM_001257138	Multisystem autoimmune disease with facial dysmorphism and autoimmune diabetes	613385	Recessive	Lohr <i>et al</i> 2010 Am J Hum Genet 86 : 447-453
JAK1 147795	NM_002227	Immune dysregulatory and hypereosinophilic syndrome	Not yet assigned	Dominant	Del Bel <i>et al</i> 2017 J Allergy Clin Immunol 139 : 2016-2020
KCNJ11 600937	NM_000525	Permanent neonatal diabetes	606176	Dominant (often <i>de novo</i>)	Gloyn <i>et al</i> 2004 N Engl J Med 350 : 1838-1849
		Transient neonatal diabetes	610582	Dominant (often <i>de novo</i>)	Yorifuji <i>et al</i> 2005 J Clin Endocrinol Metab 90 : 3174-3178 Gloyn <i>et al</i> 2005 Hum Mol Genet 14 : 925-934 Suzuki <i>et al</i> 2007 J Clin Endocrinol Metab 92 : 3979-3985
		MODY	616329	Dominant	Yorifuji <i>et al</i> 2005 J Clin Endocrinol Metab 90 : 3174-3178

					Bonnefond <i>et al</i> 2012 PLoS One 7 : e37423
LIPE 151750	NM_005357	Familial Partial Lipodystrophy (FPLD6) and insulin resistance	615980	Recessive	Farhan <i>et al</i> 2014 Canad J Cardiol 30 : 1649-1654 Zolotov <i>et al</i> 2017 Am J Med Genet 173A : 190-194
LMNA 150330	NM_170707	Familial Partial Lipodystrophy (FPLD2) and insulin resistance	151660	Dominant	Cao <i>et al</i> 2000 Hum Mol Genet 1 : 109-112 Shackleton <i>et al</i> 2000 Nat Genet 24 : 153-156 Speckman <i>et al</i> 2000 Am J Hum Genet 66 : 1192-1198
LPL 609708	NM_00237	Lipoprotein lipase deficiency and transient neonatal diabetes	238600	Recessive	Raupp <i>et al</i> 2002 J Inherit Metab Dis 25 : 413-414
LRBA 606453	NM_001199282	Immunodysregulation and autoimmune diabetes	606453	Recessive	Charbonnier <i>et al</i> 2015 J Allergy Clin Immunol 135 : 217-227 Schreiner <i>et al</i> 2016 J Clin Endocrinol Metab 101 : 898-904
MANF 601916	NM_006010	Diabetes & microcephaly	Not yet assigned	Recessive	Yavarna <i>et al</i> 2015 Hum Genet 134 : 967-980
MXN1 142994	NM_005515	Neonatal diabetes & IUGR	Not yet assigned	Recessive	Flanagan <i>et al</i> 2014 Cell Metab 19 : 146-154
MTTL1 m.3243A>G 590050	NC_012920	Maternally inherited diabetes and deafness (MIDD)	520000	Mitochondrial	Van den Ouweland <i>et al</i> 1992 Nat Genet 1 : 368-371 Murphy <i>et al</i> 2008 Diabet Med 25 : 383-399
NEUROD1 601724	NM_002500	Permanent neonatal diabetes and neurological abnormalities	Not yet assigned	Recessive	Rubio-Cabezas <i>et al</i> 2010 Diabetes 162 : 987-992
		MODY	606394	Dominant	Malecki <i>et al</i> 1999 Nat Genet 23 : 323-328
NEUROG3 604882	NM_020999	Permanent neonatal diabetes with congenital malabsorptive diarrhoea	610370	Recessive	Rubio-Cabezas <i>et al</i> 2011 Diabetes 60 : 1349-1353
NFKB1 164011	NM_003998	Immunodeficiency and autoimmunity	616576	Dominant	Fliegau <i>et al</i> 2015 Am J Hum Genet 97 : 389-403 Schröder <i>et al</i> 2019 Front Immunol 14 : 2618
NKX2-2 604612	NM_002509	Neonatal diabetes and developmental delay	Not yet assigned	Recessive	Flanagan <i>et al</i> 2014 Cell Metab 19 : 146-154
PAX6 607108	NM_001604	Aniridia and impaired glucose tolerance	106210	Dominant	Yasuda <i>et al</i> 2002 Diabetes 51 : 224-230 Nishi <i>et al</i> 2005 Diabet Med 22 : 641-644 Osawa <i>et al</i> 2015 J Diabetes Investig 6 : 105-106

PCBD1 126090	NM_000281	Diabetes and hyperphenylalaninaemia	264070	Recessive	Simaite <i>et al</i> 2014 Diabetes <u>63</u> : 3557-3564 Ferre <i>et al</i> 2014 J Am Soc Nephrol <u>25</u> : 574-586
PDX1 600733	NM_000209	Permanent neonatal diabetes +/- pancreatic agenesis	260370	Recessive	Stoffers <i>et al</i> 1997 Nat Genet <u>15</u> : 106-110 Thomas <i>et al</i> 2009 Pediatr Diabetes <u>10</u> : 492-496 De Franco <i>et al</i> 2013 Diabet Med <u>30</u> : e197-200
		MODY	606392	Dominant	Stoffers <i>et al</i> 1997 Nat Genet <u>17</u> : 138-139
PIK3R1 171833	NM_181523	SHORT syndrome	269880	Dominant	Dyment <i>et al</i> 2013 Am J Hum Genet <u>93</u> : 158-166
PLIN1 170290	NM_002666	Familial Partial Lipodystrophy (FPLD4) and insulin resistance	613877	Dominant	Gandotra <i>et al</i> 2011 N Engl J Med <u>364</u> : 740-748
POLD1 174761	NM_002691	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy (MDPL) syndrome	615381	Dominant (<i>de novo</i>)	Weedon <i>et al</i> 2013 Mat Genet <u>45</u> : 947-950
PPARG 601487	NM_015869	Familial Partial Lipodystrophy (FPLD3) and insulin resistance	604367	Dominant	Agarwal <i>et al</i> 2002 J Clin Endocrinol Metab <u>1</u> : 408-411 Barroso <i>et al</i> 1999 Nature <u>402</u> : 880-883
PPP1R15B 613257	NM_032833	Juvenile-onset diabetes with microcephaly, epilepsy and intellectual disability	616817	Recessive	Abdulkarim <i>et al</i> 2015 Diabetes <u>64</u> : 3951-3962
PTF1A 607194	NM_178161	Permanent neonatal diabetes with cerebellar and pancreatic agenesis	609069	Recessive	Sellick <i>et al</i> 2004 Nat Genet <u>36</u> : 1301-1305
RFX6 612659	NM_173560	Permanent neonatal diabetes with pancreatic hypoplasia, intestinal atresia, and gallbladder aplasia or hypoplasia	601346	Recessive	Smith <i>et al</i> 2010 Nature <u>463</u> : 775-780
SIRT1 604479	NM_012238	Monogenic autoimmune diabetes	Not yet assigned	Dominant	Biason-Lauber <i>et al</i> 2013 Cell Metab. <u>17</u> : 448-455
SLC2A2 138160	NM_000340	Fanconi-Bickel syndrome	227810	Recessive	Santer <i>et al</i> 1997 Nat Genet <u>17</u> : 324-326
SLC19A2 603941	NM_006996	Thiamine responsive megaloblastic anaemia, diabetes and deafness (TRMA) syndrome	249270	Recessive	Labay <i>et al</i> 1999 Nat Genet <u>22</u> : 300-304

SLC29A3 612373	NM_018344	H syndrome & PHID syndrome	602782	Recessive	Cliffe <i>et al</i> 2009 Hum Molec Genet <u>18</u> : 2257-2265 Molho-Pessach <i>et al</i> 2008 Am J Hum Genet <u>83</u> : 529-534
STAT1 600555	NM_007315	Immunodeficiency 31C and IPEX-like phenotype	614162	Dominant	Uzel <i>et al</i> 2013 J Allergy Clin Immun <u>131</u> : 1611-1623
STAT3 102582	NM_139276	Neonatal diabetes and polyautoimmune disease	615952	Dominant	Flanagan <i>et al</i> 2014 Nat Genet <u>46</u> : 812-814
STAT5B 604260	NM_012448	Growth hormone insensitivity with immunodeficiency	245590	Recessive	Kofoed <i>et al</i> 2003 N Engl J Med <u>349</u> : 1139-147
TNFAIP3 191163	NM_001270508	Familial Behcet-Like Autoinflammatory Syndrome and autoimmune diabetes	616744	Dominant	Zhou <i>et al</i> 2016 Nat Genet <u>48</u> : 67-73
TRMT10A 616013	NM_001134665	Juvenile-onset diabetes with microcephaly, epilepsy and intellectual disability	616033	Recessive	Igoillo-Esteve <i>et al</i> 2013 PLoS Genet <u>9</u> : e1003888
WFS1 606201	NM_006005	Wolfram syndrome (Diabetes insipidus, diabetes mellitus, optic atrophy and deafness, DIDMOAD)	222300	Recessive	Inoue <i>et al</i> 1998 Nat Genet <u>20</u> : 143-148 Strom <i>et al</i> 1998 Hum Mol Genet <u>7</u> : 2021-2028
ZBTB20 606025	NM_001164342	Primrose syndrome	259050	Dominant (<i>de novo</i>)	Cordeddu <i>et al</i> 2014 Nat Genet <u>46</u> : 815-817
ZFP57 612192	NM_001109809	Transient neonatal diabetes	601410	Recessive	Mackay <i>et al</i> 2008 Nat Genet <u>40</u> : 949-951
ZMPSTE24 606480	NM_005857	Mandibuloacral dysplasia with lipodystrophy	608612	Recessive	Agarwal <i>et al</i> 2003 Hum Mol Genet <u>12</u> : 1995-2001 Ahmad <i>et al</i> 2010 Am J Med Genet A <u>152A</u> : 2703-2710