

RADIANT Genetic Testing – Diabetes Gene List

The genes RADIANT tests that are known to cause diabetes are listed below:

Gene.Symbol	OMIM.Disease.info (MIM#)
<i>ABCC8</i>	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal dominant, Autosomal recessive
<i>AGPAT2</i>	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AIRE</i>	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal dominant, Autosomal recessive
<i>AKT2</i>	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
<i>ALMS1</i>	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>APPL1</i>	{Maturity-onset diabetes of the young, type 14}, 616511 (3), Autosomal dominant
<i>BSCL2</i>	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
<i>CEL</i>	Maturity-onset diabetes of the young, type VIII, 609812 (3), Autosomal dominant
<i>CISD2</i>	Wolfram syndrome 2, 604928 (3), Autosomal recessive
<i>CNOT1</i>	Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
<i>COL3A1</i>	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
<i>DCAF17</i>	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DNAJC3</i>	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
<i>DUT</i>	Bone marrow failure and diabetes mellitus syndrome, 620044
<i>DYRK1B</i>	Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant
<i>EIF2AK3</i>	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>FOXP3</i>	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
<i>GATA4</i>	Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant
<i>GATA6</i>	Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>GCK</i>	MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant

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<i>GLIS3</i>	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
<i>HFE</i>	{Porphyrria variegata, susceptibility to}, 176200 (3), Autosomal dominant; {Microvascular complications of diabetes 7}, 612635 (3); Hemochromatosis, 235200 (3), Autosomal recessive; {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3); {Porphyrria cutanea tarda, susceptibility to}, 176100 (3), Autosomal dominant, Autosomal recessive
<i>HNF1A</i>	Hepatic adenoma, somatic, 142330 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; MODY, type III, 600496 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; Renal cell carcinoma, 144700 (3)
<i>HNF1B</i>	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
<i>HNF4A</i>	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
<i>IER3IP1</i>	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
<i>IL2RA</i>	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
<i>INS</i>	Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 4, 618858 (3), Autosomal dominant, Autosomal recessive
<i>INSR</i>	Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Leprechaunism, 246200 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant
<i>KCNJ11</i>	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal dominant, Autosomal recessive
<i>KCNQ1</i>	Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive
<i>LIPE</i>	Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive
<i>LMNA</i>	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular

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	dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
<i>LRBA</i>	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
<i>MAX</i>	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>MC4R</i>	Obesity (BMIQ20), 618406 (3), Autosomal dominant, Autosomal recessive; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal dominant, Autosomal recessive
<i>MEN1</i>	Lipoma, somatic (3); Angiofibroma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Carcinoid tumor of lung (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3)
<i>MNX1</i>	Currarino syndrome, 176450 (3), Autosomal dominant
<i>NEUROD1</i>	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
<i>NEUROG3</i>	Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive
<i>NKX2-2</i>	
<i>PAX6</i>	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
<i>PCBD1</i>	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
<i>PDX1</i>	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
<i>PIK3R1</i>	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
<i>PLIN1</i>	Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant
<i>POLD1</i>	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
<i>PPARG</i>	{Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; [Obesity, resistance to] (3); Obesity, severe, 601665 (3), Autosomal dominant, Autosomal recessive, Multifactorial; Carotid intimal medial thickness 1, 609338 (3)
<i>PPP1R15B</i>	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
<i>PTF1A</i>	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive

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<i>RET</i>	{Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant
<i>RFX6</i>	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
<i>SDHAF2</i>	Paragangliomas 2, 601650 (3), Autosomal dominant
<i>SDHB</i>	Paragangliomas 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Pheochromocytoma, 171300 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
<i>SDHC</i>	Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases
<i>SDHD</i>	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SLC19A2</i>	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
<i>SLC29A3</i>	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC2A2</i>	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>STAT3</i>	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
<i>TMEM127</i>	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>TNNT2</i>	Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant
<i>TRMT10A</i>	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
<i>VHL</i>	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
<i>WFS1</i>	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
<i>YIPF5</i>	Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive
<i>ZBTB20</i>	Primrose syndrome, 259050 (3), Autosomal dominant
<i>ZFP57</i>	Diabetes mellitus, transient neonatal 1, 601410 (3), Autosomal dominant
<i>ZMPSTE24</i>	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive