International Textbook of Diabetes Mellitus, 4th Ed., Chapter 28 - Monogenic Disorders of the Beta Cell

References

1. Shields BM, McDonald TJ, Ellard S, et al.: The development and validation of a clinical prediction model to determine the probability of MODY in patients with young-onset diabetes. Diabetologia 2012;55(5):1265–1272.
2. Shields BM, Hicks S, Shepherd MH, et al.: Maturity-onset diabetes of the young (MODY): how many cases are we missing? Diabetologia 2010;53(12):2504–2508.
3. Pearson ER, Velho G, Clark P, et al.: Beta-cell genes and diabetes: quantitative and qualitative differences in the pathophysiology of hepatic nuclear factor-1alpha and glucokinase mutations. Diabetes 2001;50(Suppl 1):S101–107.
4. Matschinsky FM: Glucokinase, glucose homeostasis, and diabetes mellitus. Current Diabetes Reports 2005;5(3):171–176.
5. Vionnet N, Stoffel M, Takeda J, et al.: Nonsense mutation in the glucokinase gene causes early-onset non-insulin-dependent diabetes mellitus. Nature 1992;356(6371):721–722.
6. StoffelM, Froguel P, Takeda J, et al.:Human glucokinase gene: isolation, characterization, and identification of two missense mutations linked to early-onset non-insulin-dependent (type 2) diabetes mellitus. PNAS 1992;89(16):7698–7702.
7. Garcia-Herrero CM, Rubio-Cabezas O, Azriel S, et al.: Functional characterization of MODY2 mutations highlights the importance of the fine-tuning of glucokinase and its role in glucose sensing. PLoS ONE 2012;7(1):e30518.
8. Gidh-Jain M, Takeda J, Xu LZ, et al.: Glucokinase mutations associated with non-insulin-dependent (type 2) diabetes mellitus have decreased enzymatic activity: implications for structure/function relationships. PNAS 1993;90(5):1932–1936.
9. Burke CV, Buettger CW,Davis EA, et al.: Cell-biological assessment of human glucokinase mutants causing maturity-onset diabetes of the young type 2 (MODY-2) or glucokinase-linked hyperinsulinaemia (GK-HI). Biochemical Journal 1999;342(Pt 2):345–352.
10. Liang Y, Kesavan P, Wang LQ, et al.: Variable effects of maturity onset-diabetes-of-youth (MODY)-associated glucokinase mutations on substrate interactions and stability of the enzyme. Biochemical Journal 1995;309(Pt 1):167–173.
11. Njolstad PR, Sagen JV, Bjorkhaug L, et al.: Permanent neonatal diabetes caused by glucokinase deficiency: inborn error of the glucose-insulin signaling pathway. Diabetes 2003;52(11):2854– 2860.
12. Glaser B, Kesavan P, Heyman M, et al.: Familial hyperinsulinism caused by an activating glucokinase mutation. New England Journal of Medicine 1998;338(4):226–230.
13. Steele, AM, Shields BM, Wensley KJ, et al.: Prevalence of Vascular Complications Among Patients With Glucokinase Mutations and Prolonged, Mild Hyperglycemia. JAMA 2014;311(3):279–286.
14. Byrne MM, Sturis J, Clement K, et al.: Insulin secretory abnormalities in subjects with hyperglycemia due to glucokinase mutations. Journal of Clinical Investigation 1994;93(3):1120–1130.

15. Stride A, Vaxillaire M, Tuomi T, et al.: The genetic abnormality in the beta cell determines the response to an oral glucose load. Diabetologia 2002;45(3):427–435.
16. Froguel P, ZoualiH,Vionnet N, et al.: Familial hyperglycemia due to mutations in glucokinase.Definition of a subtype of diabetes mellitus. New England Journal of Medicine 1993;328(10):697–702.
17. Colom C, Corcoy R:Maturity onset diabetes of the young and pregnancy. Best Practice & Research Clinical Endocrinology & Metabolism 2010;24(4):605–615.
18. Spyer G, Macleod KM, Shepherd M, et al.: Pregnancy outcome in patients with raised blood glucose due to a heterozygous glucokinase gene mutation. Diabetic Medicine 2009;26(1):14–18.
19. Spyer G, Hattersley AT, Sykes JE, et al.: Influence of maternal and fetal glucokinase mutations in gestational diabetes. American Journal of Obstetrics & Gynecology 2001;185(1):240–241.
20. Yamagata K, Oda N, Kaisaki PJ, et al.: Mutations in the hepatocyte nuclear factor-1alpha gene in maturity-onset diabetes of the young (MODY3). Nature 1996;384(6608):455–458.
21. Wang H, Antinozzi PA, Hagenfeldt KA, et al.: Molecular targets of a human HNF1 alpha mutation responsible for pancreatic beta-cell dysfunction. EMBO Journal 2000;19(16):4257–4264.
22. Shih DQ, Screenan S, Munoz KN, et al.: Loss of HNF-1 alpha function in mice leads to abnormal expression of genes involved in pancreatic islet development and metabolism. Diabetes 2001;50(11):2472–2480.
23. Ellard S, Colclough K: Mutations in the genes encoding the transcription factors hepatocyte nuclear factor 1 alpha (HNF1A) and 4 alpha (HNF4A) in maturity-onset diabetes of the young. Human Mutation 2006;27(9):854–869.
24. Stoffers DA, Stanojevic V, Habener JF: Insulin promoter factor-1 gene mutation linked to early-onset type 2 diabetes mellitus directs expression of a dominant negative isoprotein. Journal of Clinical Investigation 1998;102(1):232–241.
25. Kristinsson SY,Thorolfsdottir ET, Talseth B, et al.:MODY in Iceland is associated with mutations in HNF-1alpha and a novel mutation in NeuroD1. Diabetologia 2001;44(11):2098–2103.
26. Liu L, Furuta H, Minami A, et al.: A novel mutation, Ser159Pro in the NeuroD1/BETA2 gene contributes to the development of diabetes in a Chinese potential MODY family. Molecular and Cellular Biochemistry 2007;303(1–2):115–120.
27. MaleckiMT, Jhala US, Antonellis A, et al.:Mutations in NEUROD1 are associated with the development of type 2 diabetes mellitus. Nature Genetics 1999;23(3):323–328.
28. PlengvidhyaN,Kooptiwut S, SongtaweeN, et al.: PAX4 mutations in Thais with maturity onset diabetes of the young. Journal of Clinical Endocrinology & Metabolism 2007;92(7):2821–2826.
29. Neve B, Fernandez-Zapico ME, Ashkenazi-Katalan V, et al.: Role of transcription factor KLF11 and its diabetes-associated gene variants in pancreatic beta cell function. PNAS 2005;102(13):4807–4812.
30. Dukes ID, Sreenan S, Roe MW, et al.: Defective pancreatic beta-cell glycolytic signaling in hepatocyte nuclear factor-1alpha-deficient mice. Journal of Biological Chemistry 1998;273(38):24457–24464.
31. Yamagata K, Nammo T, Moriwaki M, et al.: Overexpression of dominant-negative mutant hepatocyte nuclear factor-1 alpha in pancreatic beta-cells causes

abnormal islet architecture with decreased expression of E-cadherin, reduced beta-cell proliferation, and diabetes. Diabetes 2002;51(1):114–123.
32. Ferrer J: A genetic switch in pancreatic beta-cells: implications for differentiation and haploinsufficiency. Diabetes 2002;51(8):2355–2362.
33. Byrne MM, Sturis J, Menzel S, et al.: Altered insulin secretory responses to glucose in diabetic and nondiabetic subjects with mutations in the diabetes susceptibility gene MODY3 on chromosome 12. Diabetes 1996;45(11):1503–1510.
34. Byrne MM, Sturis J, Fajans SS, et al.: Altered insulin secretory responses to glucose in subjects with a mutation in the MODY1 gene on chromosome 20. Diabetes 1995;44(6):699–704.
35. Hagenfeldt-Johansson KA, Herrera PL, Wang H, et al.: Beta cell- targeted expression of a dominant-negative hepatocyte nuclear factor-1 alpha induces a maturity-onset diabetes of the young (MODY)3-like phenotype in transgenic mice. Endocrinology 2001;142(12):5311–5320.
36. Murphy R, Ellard S, Hattersley AT: Clinical implications of a molecular genetic classification of monogenic beta-cell diabetes. Nature Clinical Practice Endocrinology & Metabolism 2008;4(4):200–213.
37. Steele AM, Shields BM, Shepherd M, et al.: Increased all-cause and cardiovascular mortality in monogenic diabetes as a result of mutations in the HNF1A gene. Diabetic Medicine 2010;27(2):157–161.
38. Isomaa B, Henricsson M, Lehto M, et al.: Chronic diabetic complications in patients with MODY3 diabetes. Diabetologia 1998;41(4):467–473.
39. Pearson ER, Boj SF, SteeleAM, et al.: Macrosomia and hyperinsulinaemic hypoglycaemia in patients with heterozygous mutations in the HNF4A gene. PLoS Medicine 2007;4(4):e118.
40. Lehto M, Bitzen PO, Isomaa B, et al.: Mutation in the HNF-4 alpha gene affects insulin secretion and triglyceride metabolism. Diabetes 1999;48(2):423–425.
41. Shih DQ, Dansky HM, Fleisher M, et al.: Genotype/phenotype relationships in HNF-4 alpha/MODY1: haploinsufficiency is associated with reduced apolipoprotein (AII), apolipoprotein (CIII), lipoprotein(a), and triglyceride levels. Diabetes 2000;49(5):832–837.
42. Besser RE, Shepherd MH, McDonald TJ, et al.: Urinary C-peptide creatinine ratio is a practical outpatient tool for identifying hepatocyte nuclear factor 1-{alpha}/hepatocyte nuclear factor 4-{alpha} maturity-onset diabetes of the young from long-duration type 1 diabetes. Diabetes Care 2011;34(2):286–291.
43. Pearson ER, Starkey BJ, Powell RJ, et al.: Genetic cause of hyperglycaemia and response to treatment in diabetes. Lancet 2003;362(9392):1275–1281.
44. Tuomi T,Honkanen EH, Isomaa B, et al.: Improved prandial glucose control with lower risk of hypoglycemia with nateglinide than with glibenclamide in patients with maturity-onset diabetes of the young type 3. Diabetes Care 2006;29(2):189–194.
45. Shepherd M, Shields B, Ellard S, et al.: A genetic diagnosis of HNF1A diabetes alters treatment and improves glycaemic control in the majority of insulin-treated patients. Diabetic Medicine 2009;26(4):437–441.
46. Kapoor RR, Locke J, Colclough K, et al.: Persistent hyperinsulinemic hypoglycemia and maturity-onset diabetes of the young due to heterozygous HNF4A mutations. Diabetes 2008;57(6):1659–1663.
47. Senniappan S, Shanti B, James C, Hussain K: Hyperinsulinaemic hypoglycaemia: genetic mechanisms, diagnosis and management. Journal of Inherited Metabolic Disease 2012;35(4):589–601.

48. Rubio-Cabezas O, Klupa T, Malecki MT: Permanent neonatal diabetes mellitus—the importance of diabetes differential diagnosis in neonates and infants. European Journal of Clinical Investigation 2011;41(3):323–333.
49. Iafusco D, Stazi MA, Cotichini R, et al.: Permanent diabetes mellitus in the first year of life. Diabetologia 2002;45(6):798–804.
50. Edghill EL, Dix RJ, Flanagan SE, et al.: HLA genotyping supports a nonautoimmune etiology in patients diagnosed with diabetes under the age of 6 months. Diabetes 2006;55(6):1895–1898.
51. Stoy J, Greeley SA, Paz VP, et al.: Diagnosis and treatment of neonatal diabetes: a United States experience. Pediatric Diabetes 2008;9(5):450–459.
52. Flanagan SE, Edghill EL, Gloyn AL, et al.: Mutations in KCNJ11, which encodes Kir6.2, are a common cause of diabetes diagnosed in the first 6 months of life, with the phenotype determined by genotype. Diabetologia 2006;49(6):1190–1197.
53. Flanagan SE, Patch AM, Mackay DJ, et al.: Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. Diabetes 2007;56(7):1930–1937.
54. Gloyn AL, Pearson ER, Antcliff JF, et al.: Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. New England Journal of Medicine 2004;350(18):1838–1849.
55. Proks P, Arnold AL, Bruining J, et al.: A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. Human Molecular Genetics 2006;15(11):1793–1800.
56. Babenko AP, Polak M, Cave H, et al.: Activating mutations in the ABCC8 gene in neonatal diabetes mellitus. New England Journal of Medicine 2006;355(5):456–466.
57. Ellard S, Flanagan SE, Girard CA, et al.: Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. American Journal of Human Genetics 2007;81(2):375–382.
58. Hattersley AT, Ashcroft FM: Activating mutations in Kir6.2 and neonatal diabetes: new clinical syndromes, new scientific insights, and new therapy. Diabetes 2005;54(9):2503–2513.
59. Edghill EL, Flanagan SE, Patch AM, et al.: Insulin mutation screening in 1,044 patients with diabetes: mutations in the INS gene are a Common cause of neonatal diabetes but a rare cause of diabetes diagnosed in childhood or adulthood. Diabetes 2008;57(4):1034–1042.
60. Rubio-Cabezas O, Patch AM, Minton JA, et al.: Wolcott-Rallison syndrome is the most common genetic cause of permanent neonatal diabetes in consanguineous families. Journal of Clinical Endocrinology & Metabolism 2009;94(11):4162–4170.
61. Aguilar-Bryan L, Bryan J: Neonatal diabetes mellitus. Endocrine Reviews 2008;29(3):265–291.
62. Proks P, Antcliff JF, Lippiat J, et al.: Molecular basis of Kir6.2 mutations associated with neonatal diabetes or neonatal diabetes plus neurological features. PNAS 2004;101(50):17539–17544.
63. Stoy J, Edghill EL, Flanagan SE, et al.: Insulin gene mutations as a cause of permanent neonatal diabetes. PNAS 2007;104(38):15040–15044.
64. Julier C, Nicolino M:Wolcott-Rallison syndrome. Orphanet Journal of Rare Diseases 2010;5:29.

65. Pearson ER, Flechtner I,Njolstad PR, et al.: Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. New England Journal of Medicine 2006;355(5):467–477.
66. Koster JC, Cadario F, Peruzzi C, et al.: The G53D mutation in Kir6.2 (KCNJ11) is associated with neonatal diabetes and motor dysfunction in adulthood that is improved with sulfonylurea therapy. Journal of Clinical Endocrinology & Metabolism 2008;93(3):1054–1061.
67. Sagen JV, Raeder H, Hathout E, et al.: Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. Diabetes 2004;53(10):2713–2718.
68. Slingerland AS, Hurkx W,Noordam K, et al.: Sulphonylurea therapy improves cognition in a patient with the V59M KCNJ11 mutation. Diabetic Medicine 2008;25(3):277–281.
69. Mlynarski W, Tarasov AI, Gach A, et al.: Sulfonylurea improves CNS function in a case of intermediate DEND syndrome caused by a mutation in KCNJ11. Nature Clinical Practice Neurology 2007;3(11):640–645.
70. Edghill EL, Gloyn AL, Goriely A, et al.: Origin of de novo KCNJ11 mutations and risk of neonatal diabetes for subsequent siblings. Journal of Clinical Endocrinology & Metabolism 2007;92(5): 1773–1777.
71. Temple IK, Gardner RJ, Mackay DJ, et al.: Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. Diabetes 2000;49(8):1359–1366.
72. Mackay DJ, Callaway JL, Marks SM, et al.: Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. Nature Genetics 2008;40(8):949–951.
73. Gloyn AL, Reimann F, Girard C, et al.: Relapsing diabetes can result from moderately activating mutations in KCNJ11. Human Molecular Genetics 2005;14(7):925–934.
74. Yorifuji T, Nagashima K, Kurokawa K, et al.: The C42R mutation in the Kir6.2 (KCNJ11) gene as a cause of transient neonatal diabetes, childhood diabetes, or later-onset, apparently type 2 diabetes mellitus. Journal of Clinical Endocrinology & Metabolism 2005;90(6):3174–3178.
75. Colombo C, Delvecchio M, Zecchino C, et al.: Transient neonatal diabetes mellitus is associated with a recurrent (R201H) KCNJ11 (KIR6.2) mutation. Diabetologia 2005;48(11):2439–2441.
76. Patch AM, Flanagan SE, Boustred C, et al.:Mutations in the ABCC8 gene encoding the SUR1 subunit of the KATP channel cause transient neonatal diabetes, permanent neonatal diabetes or permanent diabetes diagnosed outside the neonatal period. Diabetes, Obesity and Metabolism 2007;9(Suppl 2):28–39.
77. Temple IK, Shield JP: Transient neonatal diabetes, a disorder of imprinting. Journal of Medical Genetics 2002;39(12):872–875.
78. Bingham C, Bulman MP,EllardS, et al.:Mutations in the hepatocyte nuclear factor-1beta gene are associated with familial hypoplastic glomerulocystic kidney disease. American Journal of Human Genetics 2001;68(1):219–224.
79. Bingham C, Ellard S, Cole TR, et al.: Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1 beta mutations. Kidney International 2002;61(4):1243–1251.
80. Coffinier C, Barra J, Babinet C, Yaniv M: Expression of the vHNF1/HNF1beta homeoprotein gene during mouse organogenesis. Mechanisms of Development 1999;89(1–2):211–213.

81. Edghill EL, Bingham C, Slingerland AS, et al.: Hepatocyte nuclear factor-1 beta mutations cause neonatal diabetes and intrauterine growth retardation: support for a critical role of HNF-1beta in human pancreatic development. Diabetic Medicine 2006;23(12):1301–1306.
82. Haldorsen IS, Vesterhus M, Raeder H, et al.: Lack of pancreatic body and tail in HNF1B mutation carriers. Diabetic Medicine 2008;25(7):782–787.
83. Bingham C, Ellard S, Allen L, et al.: Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1 beta. Kidney International 2000;57(3):898–907.
84. Heidet L, Decramer S, Pawtowski A, et al.: Spectrum of HNF1B mutations in a large cohort of patients who harbor renal diseases. Clinical Journal of the American Society of Nephrology 2010;5(6):1079–1090.
85. Adalat S, Woolf AS, Johnstone KA, et al.: HNF1B mutations associate with hypomagnesemia and renal magnesium wasting. Journal of the American Society of Nephrology 2009;20(5):1123–1131.
86. Bingham C, Ellard S, van't Hoff WG, et al.: Atypical familial juvenile hyperuricemic nephropathy associated with a hepatocyte nuclear factor-1beta gene mutation. Kidney International 2003;63(5):1645–1651.
87. Bellanne-Chantelot C, Chauveau D, Gautier JF, et al.: Clinical spectrum associated with hepatocyte nuclear factor-1 beta mutations. Annals of Internal Medicine 2004;140(7):510–517.
88. Moreno-De-Luca D, Mulle JG, Kaminsky EB, et al.: Deletion 17q12 is a recurrent copy number variant that confers high risk of autism and schizophrenia. American Journal of Human Genetics 2010;87(5):618–630.
89. Alcolado JC, Laji K, Gill-Randall R: Maternal transmission of diabetes. Diabetic Medicine 2002;19(2):89–98.
90. van den Ouweland JM, Bruining GJ, Lindhout D, et al.: Mutations in mitochondrial tRNA genes: non-linkage with syndromes of Wolfram and chronic progressive external ophthalmoplegia. Nucleic Acids Research 1992;20(4):679–682.
91. Kadowaki T,Kadowaki H,Mori Y, et al.:A subtype of diabetes mellitus associated with a mutation of mitochondrial DNA. New England Journal of Medicine 1994;330(14):962–968.
92. Maassen JA, Kadowaki T: Maternally inherited diabetes and deafness: a new diabetes subtype. Diabetologia 1996;39(4):375–382.
93. Murphy R, Turnbull DM, Walker M, Hattersley AT: Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. Diabetic Medicine 2008;25(4):383–399.
94. Barrett TG, Bundey SE, Macleod AF: Neurodegeneration and diabetes: UK nationwide study of Wolfram (DIDMOAD) syndrome. Lancet 1995;346(8988):1458–1463.
95. Khanim F, Kirk J, Latif F, Barrett TG: WFS1/wolframin mutations, Wolfram syndrome, and associated diseases. Human Mutation 2001;17(5):357–367.
96. Amr S,Heisey C, Zhang M, et al.:Ahomozygous mutation in a novel zinc-finger protein, ERIS, is responsible for Wolfram syndrome 2. American Journal of Human Genetics 2007;81(4):673–683.
97. Chaussenot A, Bannwarth S, Rouzier C, et al.: Neurologic features and genotype-phenotype correlation in Wolfram syndrome. Annals of Neurology 2011;69(3):501–508.

98. Ellard S, Bellanne-Chantelot C, Hattersley AT: Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. Diabetologia 2008;51(4):546–553.