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Appendix

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Appendix to: Bishay RH, Greenfield JR. A review of maturity onset diabetes of the young (MODY) and challenges in the management of glucokinase-MODY. *Med J Aust* 2016; 205: 480-485. doi: 10.5694/mja16.00458.

Appendix. Prevalence, genetic and key clinical features of the common forms of maturity onset diabetes of the young

MODY subtype	Prevalence (% estimate of all MODY subtypes)	Mutation	Clinical features	Diagnostic glucose and HbA_{1c}	Treatment	Special features
1	~ 5%	*HNF4A*[1]	Young age (< 25 years); strong family history of diabetes; absent antibodies; detectable C-peptide	As with type 2 diabetes; postprandial glucose excursions, ≥ 5 mmol	Sensitive to SU; 30–40% apparent insulin-requiring	Glycosuria common; develop micro- and macrovascular complications as in type 1 and 2 diabetes
2	10–60%	*GCK*[2]		Fasting BGL, 5.4–8.3 mmol/L; post-prandial glucose excursions, ≤ 3mmol/L; HbA_{1c}, 5.8–7.6% (40–60 mmol/mol)	None required (controversial)	Favourable lipid profile; lean; minimal or no micro or macrovascular complications; minimal affect of treatment on glycaemic control
3	20–50%	*HNF1A*[3,4]		As for MODY1	As for MODY1	As for MODY1; strong family history of macrosomic babies
4	< 1%	*PDX1 (IPF1)*[5]	Average onset 35-years; pancreatic agenesis in homozygous and compound heterozygotes			
5	~ 5%	*HNF1B (TCF2)*[6-8]	"Renal cysts and diabetes syndrome;" female genital malformations, hyperuricaemia, pancreatic atrophy, abnormal liver function tests			
6	Rare < 1%	*NEUROD1*[5]	Very rare; adult onset (mid-20s); reduced insulin production due to developmental β-cell dysfunction; overweight/obese, similar to type 2 diabetes mellitus			
7		*KLF11*[9]	Heterozygous mutation; early adult onset diabetes			
8		*CEL*[10]	Adult onset (mean age 36 years); exocrine pancreatic insufficiency with low faecal elastase			
9		*PAX4*[11]	Adult onset (29–49 years), may be seen in ketosis-prone diabetes; may need insulin			
10		*INS*[12]	Variable presentation; usually neonatal diabetes, but affects early childhood and adults (15–65 years)			
11		*BLK*[13]	Mean age (31 years), normal/overweight (BMI, 28 kg/m^2); 60% require insulin; high fasting and postprandial BGLs			
12		*ABCC8*[14]	Similar to MODY1 and MODY3			
13		*KCNJ11*[15]	Ages 3–26 years, neonatal diabetes, prediabetes or childhood or adult-onset diabetes; respond to SU			
14		*APPL1*[16]	Adult onset diabetes (35–48 years); loss-of-function mutation.			

ABCC8 = ATP-binding cassette transporter subfamily c member 8. *APPL* = adaptor protein, phosphotyrosine interaction, PH domain, and leucine zipper containing 1. *BLK* = B-lymphocyte kinase *(BLK) I. CEL* = carboxyl-ester lipase. *GCK* = glucokinase. *HNF1A* = hepatocyte nuclear factor 1α. *HNF4A* = hepatocyte nuclear factor 4α. *HNF1B* = hepatocyte nuclear factor 1β. *INS* = insulin gene. *IPF1* = insulin promoter factor 1. *KCNJ11* = potassium voltage-gated channel subfamily 11. *KLF11* = kruppel-like factor 11. *NEUROD1* = encoding neurogenic differentiation 1. *PAX4* = paired box gene 4. *PDX1* = pancreas/duodenum homeobox protein 1. SU = sulphonylurea. *TCF2* = transcription-like factor 2.

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