Annotation of genes associated with Maturity onset diabetes of the young (MODY)

Gene	Phenotype	MODY Subtype
GCK	Persistent and mild fasting hyperglycaemia from birth	MODY2
HNF1A	Common forms of MODY that can be treated with sulphonylureas	MODY3
HNF4A	Common forms of MODY that can be treated with sulphonylureas	MODY1
KCNJ11	Rare forms of MODY that can be treated with sulphonylureas	MODY13
ABCC8	Rare forms of MODY that can be treated with sulphonylureas	MODY12
HNF1B	Diabetes and exocrine dysfunction	MODY5
CEL	Diabetes and exocrine dysfunction	MODY8
NEUROD1	Rare, non-syndromic, autosomal dominant forms of MODY	MODY6
INS	Rare, non-syndromic, autosomal dominant forms of MODY	MODY10
PDX1	Rare, non-syndromic, autosomal dominant forms of MODY	MODY4
APPL1	Rare, non-syndromic, autosomal dominant forms of MODY	MODY14
PAX4		MODY9
BLK		MODY11
KLF11		MODY7
CISD2	Wolfram syndrome (diabetes, optic atrophy and deafness)	
DCAF17	Diabetes_MODY	Rare, non-syndromic, autosomal dominant forms of MODY
DNAJC3	Autosomal recessive juvenile-onset diabetes with central and peripheral neurodegeneration	
DYRK1B	Diabetes and metabolic syndrome	
GATA4	Diabetes and cardiac malformations	
GATA6	Diabetes and cardiac malformations	

INSR	Severe insulin resistance without lipodystrophy and dyslipidaemia	
LMNA	Partial lipodystrophy, dyslipidaemia and insulin resistance	
PAX6	Aniridia and diabetes	
PCBD1	Rare, non-syndromic, autosomal recessive forms of MODY	
PIK3R1	Diabetes and SHORT syndrome (short stature, hyperextensibility, hernia, ocular depression, Rieger anomaly, and teething delay)	
PLIN1	Partial lipodystrophy, dyslipidaemia and insulin resistance	
POLD1	MDPL (Mandibular hypoplasia, deafness, progeroid features and lipodystrophy) Syndrome	
PPARG	Partial lipodystrophy, dyslipidaemia and insulin resistance	
PPP1R15B	Autosomal recessive juvenile-onset diabetes with microcephaly, epilepsy and intellectual disability	
RFX6	Rare, non-syndromic, autosomal dominant forms of MODY	
SLC29A3	H syndrome (hyperpigmentation, hypertrichosis, hepatosplenomegaly, heart anomalies, hearing loss, hypogonadism, low height, and hyperglycaemia) and PHID syndrome (pigmented hypertrichosis with insulin dependent diabetes)	
TRMT10A	Autosomal recessive juvenile-onset diabetes with microcephaly, epilepsy and intellectual disability	
WFS1	Wolfram syndrome (diabetes, optic atrophy and deafness)	
ZBTB20	Primrose syndrome (tall stature, macrocephaly, intellectual disability, disturbed behaviour, unusual facial features, diabetes, deafness, progressive muscle wasting and ectopic calcifications)	
ZFP57	Rare, non-syndromic, autosomal recessive forms of MODY	