

Table 1. Some endocrine disorders for which genetic testing is available

Disease	Locus	Gene involved	Conventional diagnosis	Utility of molecular Dx
CAH	6p21	21-hydroxylase	Available, reliable	Useful for <i>in utero</i> treatment
Androgen insensitivity	Xq11	Androgen receptor	By exclusion, fairly reliable	Important to confirm in partial cases
MEN1	11q13	Menin	Only after tumors develop	Surveillance for tumors in mutation carriers
MEN2 & Familial medullary thyroid Ca	10q11.2	<i>RET</i> oncogene	Calcitonin levels, unreliable for prediction	Life-saving preventive thyroidectomy
Pseudohypoparathyroidism	20q13	<i>GNAS</i>	Available, reliable	Types 1a and 1b can be distinguished
Autoimmune polyendocrinopathy type 1	21q22.3	AIRE	Usually clear	Research only
Testotoxicosis	2p21	LH receptor	Usually clear	Research only
Hypopituitarism	Various	Various	Clear if deficiency is complete	Research only
Hyperinsulinemic hypoglycemia of infancy, recessive	11p13	<i>SUR & KIR6.2</i>	Distinction between focal and diffuse form often problematic	Distinction of focal vs. diffuse may help therapy (surgery vs. conservative)
Hyperinsulinemic hypoglycemia of infancy, dominant	10q23.3	Glutamate dehydrogenase	Hyperammonemia allows specific diagnosis	Research only
Transient neonatal diabetes	6q24	Probably <i>ZAC</i>	Clinical course characteristic	Research only
MODY1	20q12	<i>HNF4a</i>	Does not distinguish sub-types	Research only
MODY2	7p15	<i>GCK</i>	Does not distinguish sub-types	Research only
MODY3	12q24	<i>HNF1a</i>	Does not distinguish sub-types	Research only
MODY4	13q21	<i>IPF1 (PDX1)</i>	Does not distinguish sub-types	Research only
MODY5	17q21.3	<i>TCF2</i>	Does not distinguish sub-types	Renal prognosis
MODY6	2q32	<i>NEUROD1</i>	Does not distinguish sub-types	Research only
Prader-Willi	15q12	SNRPN	Based on morphological phenotype	Prognosis and life planning