Medizinische Genetik Mainz

Genetic analyses for metabolic diseases and endocrinopathies

Genetic metabolic diseases usually progress insidiously and are far more common than thought. Above all, the primarily unspecific clinical symptoms often make a fast and correct diagnosis difficult. Disorders of our endocrine system often show overlapping symptoms and are just as often genetically caused. They affect both children and adults and can occur as an isolated symptom or as part of a genetic syndrome within a more complex clinical picture. If left untreated, endocrinopathies impair physical and mental function and development and lead to severe organ damage.
With knowledge of the underlying genetic alteration, the diagnosis can be reliably confirmed, specified and sometimes corrected (so-called reverse phenotyping by modern genetic diagnostics). An early and timely diagnosis can individually optimise patients' treatment and usually avoid long-term damage. Knowledge of the exact inheritance pattern helps identify carriers in the family and determine the risk of recurrence for offspring.

A	Adrenal hyperplasia*, congenital
C	Creatine Deficiency
D	Diabetes mellitus, permanent neonatal
	Diabetes mellitus, transient neonatal
	Diabetes, monogenic
	MODY (Maturity onset diabetes of the young)
	MODY-like diseases
F	Fatty acid oxidation and ketogenesis disruption
G	Glucocorticoid deficiency
	Disorders of Gluconeogenesis
	Glycogen storage disorders
	Glycosylation disorder, congenital
	Growth disorders
H	Hypercholesterolemia
	Hyperinsulinism, congenital
	Hyperparathyroidism
	Hypertriglyceridemia (including FCS, MCS)
	Hypoglycemia
	Hypogonadotropic hypogonadism
	Hypoparathyroidism
	Hypothyroidism (including Thyroid hormone resistance)
I	Insulin deficiency, syndromic
	Insulin resistance
K	Kabuki syndrome
	Kallmann syndrom
	Ketolysis disorder

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L	Lipodystrophy
M	Mucopolysaccharidosis
O	Obesity
P	Post-INSR signaling pathway disorders
S	Sex development disorders

* Analysis provided by a group partner laboratory

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