Medizinische Genetik Mainz

Genetic analyses for imprinting disorders

In the course of human development, defined groups of genes are imprinted by specific methylation. This process is referred to as „Imprinting" and naturally leads to the inactivation of one of the two parental alleles. Depending on their parental origin, genes subjected to imprinting are inherited in an active or inactive form. Imprinting diseases are congenital and are caused by defective imprinting resulting in misregulated expression of the affected genes. Each imprinting disorder can have multiple genetic or epigenetic causes, each of which carries very different recurrence risks, differing according to parental origin. This results in an enormous complexity in genetic diagnosis. Clinically, imprinting disorders are very heterogeneous. Depending on the (epi)genetic cause, the phenotypic manifestations often include developmental delays, cognitive impairments and growth disorders. Precise molecular genetic clarification is essential for accurate diagnosis, prognosis and individualized treatment of patients.

- Angelman syndrome (AS)
- Beckwith-Wiedemann syndrome (BWS)
- Prader-Willi syndrome (PWS)
- Silver-Russel syndrome (SRS)
- Transient neonatal diabetes mellitus (TNDM)

Medizinische Genetik Mainz | Haifa-Allee 38 | 55128 Mainz
info@medgen-mainz.de | www.medgen-mainz.de | T: +49 6131 27661-0