



Uniparental Disomy (UPD) in Clinical Genetics: A Guide for Clinicians and Patients

Thomas Liehr

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This book focus on genetic diagnostics for Uniparental Disomy (UPD), a chromosomal disorder defined by the exceptional presence of a chromosome pair derived from only one parent, which leads to a group of rare diseases in humans. First the molecular and cytogenetic background of UPD is described in detail; subsequently, all available information of the various chromosomal origins and the latest findings on genotype-phenotype correlations and clinical consequences are discussed.

Numerous personal reports from families with a child suffering from a UPD-induced syndrome serve to complement the scientific and clinical aspects. Their experiences with genetic counseling and living with a family member affected by this chromosomal aberration present a vivid picture of what UPD means for its victims.

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