Biomedicine: Human Genetics

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Uniparental Disomy (UPD) in Clinical Genetics

A Guide for Clinicians and Patients

- First book on this topic
- The author is a leading expert in the field and his laboratory is collecting all published cases
- Written in collaboration with a family support group
- With numerous patient reports

This book focuses 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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