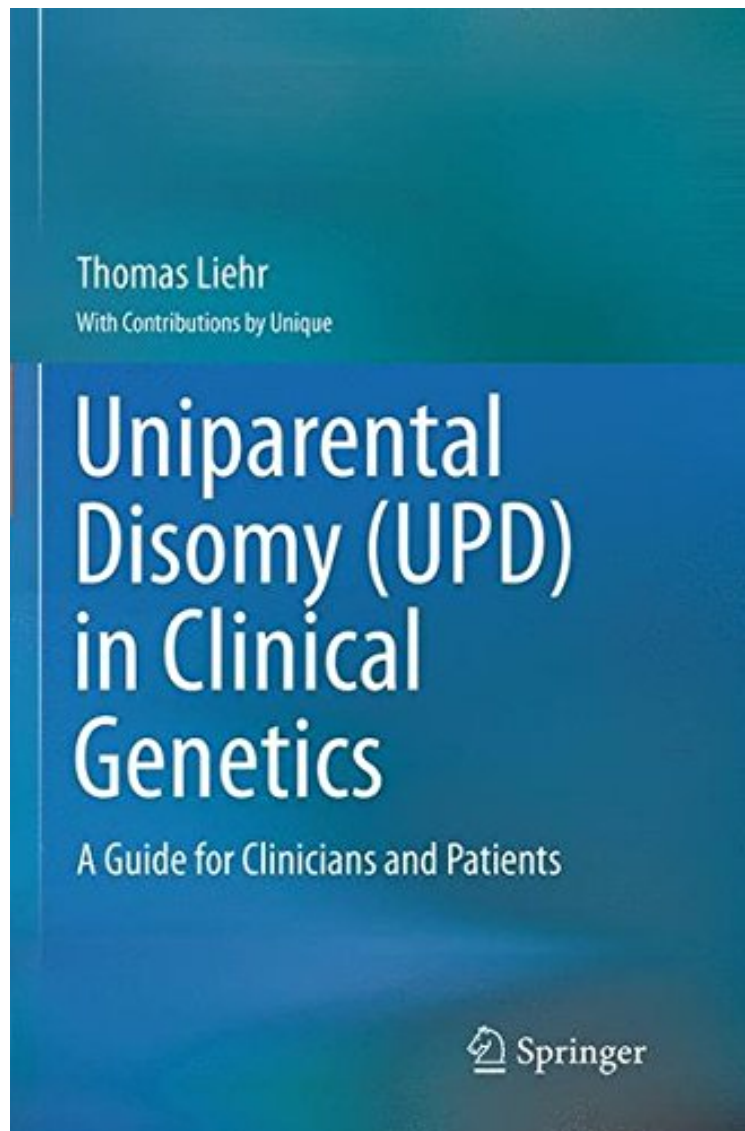


Uniparental Disomy (UPD) in Clinical Genetics: A Guide for Clinicians and Patients

Thomas Liehr

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Thomas Liehr : Uniparental Disomy (UPD) in Clinical Genetics: A Guide for Clinicians and Patients before purchasing it in order to gage whether or not it would be worth my time, and all praised Uniparental Disomy (UPD) in Clinical Genetics: A Guide for Clinicians and Patients:

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geneticists aren't familiar ...By CustomerWe have a son with MUPD14, there are very few resources out there. This is a good source of information- especially considering many geneticists aren't familiar with this under diagnosed condition so it's hard to get answers. Also, if you have a child with any UPD condition, register with The Unique Foundation (based in the UK), who will put you in touch with other parents whose children have the same syndrome if you wish. This has been the best thing we've done for our son- as we've been given great insights from other parents on early intervention, growth hormone treatment, etc. Thank you to the authors for publishing this information.

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.

From the book reviews: The book discusses basic concepts avoiding technological language, making book accessible for both lay people and clinical geneticists. This is a highly specialized book on a specific molecular/cytogenetic process. It should be a great aid for clinical geneticists to make them aware of the phenotype expected in situations where UPD is involved. Patients with UPD and their parents may find the book informative. (Luis F. Escobar, Doodys Book s, September, 2014) From the Back Cover 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.