

Preface

Uniparental disomy (UPD) is a topic, which normally is considered to be something molecular genetics has to take care of exclusively. As UPD is characterized by microsatellite analyses in the majority of the cases, which is obviously a molecular genetic approach, this assumption was never really scrutinized during the last decades.

I came across the topic UPD as a molecular cytogeneticist working on small supernumerary marker chromosomes (sSMC) (Liehr 2012 and 2014d). Besides centromere-near imbalances, mosaicism, and other factors, UPD also has to be considered in sSMC carriers exhibiting clinical problems. As reviews on UPD are scarce I started in 2010 collecting all published UPD cases in a freely available database (Liehr 2010 and 2014c) and went more and more into this topic. Surprisingly, at least for me, it turned out: UPD is a chromosomal disorder and thus it has to be a primary topic not only for molecular geneticists but also and especially for cytogeneticists. This is valid, as chromosomal alterations being detectable in $\sim 30\%$ of UPD cases, can be not only a hint on UPD presence; they also can be (and most often are) the underlying reason for UPD-formation. UPD in clinical cases most often is based on extensive, impressive, and yet not well understood capacities of human cells to correct chromosomal imbalances and/or rearrangements. UPD provides otherwise impossible insights in these repair capacities, which appear during gametogenesis and early embryogenesis. Most interesting, there is also so-called acquired UPD, which seems to be of tremendous relevance in tumor progression.

As UPD leads to a group of rare diseases in human, this book has not only the goal to collect and present the yet available information on UPD; besides also patients carrying a UPD and/or families having a child with a UPD-induced syndrome report their experiences with diagnostics, counseling and living with such a syndrome. Only the inclusion of these family reports provides a most comprehensive picture of what UPD really means.

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A Guide for Clinicians and Patients

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