Abstract

Chromosome pairs affect how our body works. Normally, a baby gets one copy of each chromosome from each parent. This means one copy from mother, and the other copy from father. In rare cases, a baby may get two copies from the same parent and none from the other. This phenomenon is called uniparental disomy (UPD). Uniparental disomy is a major topic for molecular geneticists and cytogeneticists. There are several mechanisms that lead to UPD, for example: gamete complementation, monosomy rescue, trisomy rescue or post-fertilization error. The consequences of UPD can be diverse and depend on the specific chromosome and genetic content of the affected region. An abnormal phenotype is manifested if the UPD occurs on a chromosome that is subject to genomic imprinting. An abnormal phenotype can also occur due to mutations. Among the most common syndromes associated with UPD are Prader-Willi syndrome and Angelman syndrome, which I focus on in my theses. I consider the greatest contribution of my theses to be the creation of a comprehensive overview of the effects of UPD across all human chromosomes.