Hypophosphatasia due to uniparental disomy.

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Abstract

We have read with interest the paper by Watanabe et al. [1] describing a fetus with hypophosphatasia (HPP) caused by paternal uniparental disomy (UPD) of chromosome 1 resulting in homozygosity for an ALPL mutation. Although we have recently identified a similar case, we are not convinced that the claim by Watanabe et al. that UPD can be a frequent mechanism of recessive disorders is supported by current data. However, it should be stressed that UPD is not the only reason why parental genotypes should always be tested when homozygosity for a recessive mutation is identified in the child, especially in a prenatal diagnostic setting.


Published: 31. 8. 2016 / Responsible person: Mgr. Ing. Tereza Kůstková

Source URL (retrieved on 26. 10. 2018 - 14:21):