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Titel des Beitrags: Cryptic del/dup aberration of 60.6 Mb at 5q15-5q23.3 predicting adult-onset leukodystrophy.

Abstract: We report on a de novo interstitial del/dup aberration consisting of a 13.3 Mb deletion of 5q15-5q21.3 (92.1-105.4 Mb, hg19) and a 23.6 Mb tandem direct duplication of 5q21.3-5q23.3 (106.1-129.7 Mb, hg19). Although the aberration covered a total of 60.6 Mb, it was cryptic, i.e., not detectable by karyotyping at a resolution of 430 bands. Array-CGH indicated a diploid region of 0.6 Mb between the duplicated and the deleted segment. The aberration affected a 14-month-old boy conceived after intracytoplasmic sperm injection who presented with developmental delay, muscular hypotonia, partial agenesis of the corpus callosum, prominent forehead, low set ears, hypertelorism, hyperopia, wide-bridged nose, retrognathia, high palate, and cryptorchidism. The duplicated segment comprised the LMNB1 gene, thus predicting adult-onset autosomal-dominant leukodystrophy and revealing a temporal dimension of the phenotype. Counseling problems implicated by this prediction include "the right not to know" that the patient might want to exercise when coming of age.

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