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Supernumerary Asymmetric Dic(15;15) With Secondary Mosaic Formation in One of Two Developmentally Retarded Twins*

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ABSTRACT Two male twins born after IVF therapy were investigated for 9 years. Lymphocyte culture at the age of 1 and 9 years revealed that one twin (P1) carried a supernumerary asymmetric dic(15;15) which was present in 100% of cells. In buccal mucosa, however, the derivative 15 (der(15)) was lost in 12% of cells. The der(15) was maternal in origin and was determined to be a meiosis I abnormality. The asymmetric structure of the der(15), as demonstrated by GTG banding and microarray analysis, resulted in a tetrasomy for the region 15q11q13.2 and trisomy for 15q13.2q13.3. Both regions were interrupted by a disomic fragment of 514 kb; this region has been published as a CNV. Monosomy for this CNV was confirmed in the mother, along with a second CNV in the distal breakpoint region in 15q13.3. The patient had a second structural aberration, namely an almost complete deletion of one of the two centromeres in the der(15), such that it was not detected by metaphase or interphase FISH with the DNA probe D15Z4. The conventional karyotypes of the parents and the second twin (P2) were normal. The second twins retardation was obviously caused by an infection at the age of 4 weeks, followed by multiple organ failure. Twin P1 received special support from birth to the age of 9 years and has developed better than was to be expected from findings in the literature, while his twin brother failed to show developmental progress.