

Non-Mosaic Trisomy 7 in Chorionic Villi and Trisomy 18 in the Fetus: An Extreme form of Mosaic Variegated Aneuploidy?

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ABSTRACT Mosaic variegated aneuploidy (MVA) is characterized by multiple trisomies. The parallel existence of two non-mosaic trisomies of two different chromosomes in the fetus and the placenta has not been published to date. We report here on a putative extreme form of MVA in a pregnancy with a non-mosaic trisomy 7 in CVS and a non-mosaic trisomy 18 in amniotic fluid. The trisomy 7 was not detected in amniocytes, but a non-mosaic trisomy 18 was diagnosed. Both aneuploidies were confirmed through STR typing of the respective tissue. We infer a postzygotic mitotic origin of both aneuploidies based on the observed reduction of maternal heterozygosity to homozygosity in the analysed markers. It is indeed well conceivable that mitotic errors occur in the early embryo briefly after differentiation into trophoblast and epiblast, resulting in a complete fetal-placental discordance such as the one observed in our case. However, the observation that trisomies 7 and 18 are among the most common aberrations in MVA would support our assumption that the complete discordance of the chromosomal complement in our case represents an extreme form of MVA.