

# Marker Chromosomes and Implications for Genetic Counseling

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## Abstract/Resumo

Small supernumerary marker chromosomes (sSMCs) are chromosomal fragments or markers whose origins often requires both conventional cytogenetic methods and molecular approaches for a characterization. Microarray Chromosome Analysis (MCA) and fluorescence in situ hybridization (FISH) provide accurate characterization of sSMCs in terms of chromosomal origin, gene content and other additional imbalances elsewhere in the genome. sSMCs have been reported to be present in ~0.043% of live births and ~0.075% prenatal cases, are seven times more prevalent in intellectually disabled patients, with a frequency of 0.125% in individuals presenting infertility. Approximately 70% of SMCs are de novo and 30% are inherited, either from the mother (20%) or the father (10%). The implication of sSMC could be due to partial trisomy or tetrasomy of some genes and the effects on the phenotype is difficult to define. It depends on factors such as level of the mosaicism, size and gene content. There are genomic regions which can be present in three or more copies without causing harm, whereas other regions are dose-sensitive. If no genetically relevant material can be detected, in several cases this means that the sSMC will have no direct clinical impact on the phenotype of its carrier. Most critical are the prenatal sSMC cases. The risk depends on a number of factors, including ultrasound findings, whether the SMC is familial and if it is associated with a known syndrome. For some cases, the chromosomal abnormality led to an even more imbalanced, and subsequently unviable, situation in the potential offspring than that present in the sSMC carriers themselves. Thus, the correct identification and characterization of sSMCs has a major impact on the families.

Keyword/Palavras-chave: sSMCs; Genetic Counseling; Impact on the families