## عنوان مقاله:

Prenatal diagnosis of de novo small supernumerary marker chromosome ۴q (۴q۱۱-q۱۲): A case report

## محل انتشار:

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تعداد صفحات اصل مقاله: 6

## نویسندگان:

.Reza Mohammadi - *Genetic Laboratory of Shiraz Fertility Center, Shiraz, Iran*

.Raheleh Taheri - *Genetic Laboratory of Shiraz Fertility Center, Shiraz, Iran*

.Fatemeh Shahriyari - *Genetic Laboratory of Shiraz Fertility Center, Shiraz, Iran*

.Farnaz Feiz - *Shiraz University of Medical Sciences, Shiraz, Iran*

.Zahra Mohammadi - *Pathobiology Laboratory of Ordibehesht Hospital, Shiraz, Iran*

Sadegh Shirian - *Department of Pathology, School of Veterinary Medicine, Shahrekord University, Shahrekord, Iran. Shiraz Molecular Pathology Research Center, Dr Daneshbod Pathol Lab, Shiraz, Iran. Shefa Neurosciences Research Center, Tehran, Iran.*

.Reza Rraoofian - *Legal Medicine Research Center, Legal Medicine Organization, Tehran, Iran*

.Abdorrasoul Malekpour - *Legal Medicine Research Center, Legal Medicine Organization, Tehran, Iran*

Reza Pazhoomand - *Genetic Laboratory of Shiraz Fertility Center, Shiraz, Iran. Legal Medicine Research Center, Legal Medicine Organization, Tehran, Iran.*

## خلاصه مقاله:

Background: Small supernumerary marker chromosomes (sSMCs) are chromosomal fragments with abnormal structures found in patients with fertility problems and developmental delay. They may be detected in amniotic cell karyotypes. sSMCs are categorized as hereditary or de novo. Here, we describe a case of prenatal de novo ۴q۱۱q۱۲ sSMC and its molecular cytogenetic features which had no apparent phenotypic abnormality. Case: The fetus of a ۳۶-yr-old pregnant woman was detected positive for Down's syndrome (trisomy ۲۱) at the ۱۶th wk of gestation. Quantitative Fluorescent polymerase chain reaction technique was applied for the rapid detection of numerical aneuploidy of chromosomes X, Y, ۱۳, ۱۸, and ۲۱ microsatellites. Array comparative genomic hybridization (array CGH) technique was also conducted following the karyotype analysis of amniotic cells. The karyotype analysis was also done for the parents. Quantitative Fluorescent polymerase chain reaction result revealed a male fetus with a normal chromosomal pattern, while the amniocentesis karyotype analysis identified a male fetus with a marker chromosome (۴۷, XY, +mar), and the sSMC were existing in ۱۰۰% of amniocyte metaphase spreads. The parents' normal karyotypes indicated that the sSMC was de novo. Array CGH analysis revealed a ۶.۴۸-Mb duplication at ۴q۱۱q۱۲. Eventually, the parents decided to terminate the pregnancy by legal abortion. Conclusion: Our study highlights the importance of the application of

array CGH in combination with karyotype analysis for rapid and precise prenatal diagnosis of partial aneuploidy region.


## کلمات کلیدی:

Prenatal diagnosis, Array CGH, Chromosome ۴, Chromosome Markers., تشخیص پیش از تولد, Array CGH, کروموزوم ۴, ۴q.


## لینک ثابت مقاله در پایگاه سیویلیکا:

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