

## عنوان مقاله:

A De Novo Deletion of Chromosome 18p with Persistent Limb Tremor and Difficulty Speaking

## محل انتشار:

علوم اعصاب کاسپین، دوره 5، شماره 1 (سال: 1398)

تعداد صفحات اصل مقاله: 7

## نویسندگان:

Aghil Esmaeili-Bandboni - *Department of Medical Genetics, School of Medicine, Guilan University of Medical Sciences, Rasht, Iran*

Arash Davoudi - *Division of Cytogenetic, Dr. Keshavarz Medical Genetics Lab, Rasht, Iran*

Forozan Milani - *Reproductive Health Research Center, Alzahra Hospital, Department of Obstetrics and Gynecology, School of Medicine, Guilan University of Medical Sciences, Rasht, Iran*

Sara Afzali - *Division of Cytogenetic, Dr. Keshavarz Medical Genetics Lab, Rasht, Iran*

Alireza Sharafshah - *Division of Cytogenetic, Dr. Keshavarz Medical Genetics Lab, Rasht, Iran. Cellular and Molecular Research Center, Department of Medical Genetics, School of Medicine, Guilan University of Medical Sciences, Rasht, Iran*

Fereshteh Fallahabadi - *Division of Cytogenetic, Dr. Keshavarz Medical Genetics Lab, Rasht, Iran*

## خلاصه مقاله:

Background: The common causes of 18p deletion syndrome are spontaneous errors in the chromosomal structure in the early stages of human embryonic development. Clinical Presentation and Intervention: In this study, a 29-year-old girl was introduced with the features of deletion of chromosome 18. In addition, GTG banding karyotype revealed that this case had a deletion involving the short arm of chromosome 18. In comparison with the usual phenotype of 18p deletion, many phenotypical features of this case were similar to the other cases of 18p monosomy. Conclusion: However, two new features; difficulty in speaking and persistent limb tremor, were found that had not been observed in previous studies on the 18p deletion. Speaking was without obvious pronunciation, and the patient's physical movements were always unbalanced. These two features can be new signs for 18p deletion syndrome

## کلمات کلیدی:

Chromosome deletion, Chromosome 18, Tremor, Karyotype

## لینک ثابت مقاله در پایگاه سیویلیکا:

<https://civilica.com/doc/991166>



