GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Case Report

Molecular characterization of microduplication 22q11.2 in a girl with hypernasal speech

Y. Soysal[1], J. Vermeesch[2], N.A. Davani[2], N. Şensoy[3], K. Hekimler[1] and N. İmirzalıoğlu[1]

[1]Department of Medical Genetics, Faculty of Medicine, Afyon Kocatepe University, Afyonkarahisar, Turkey
[2]Center for Human Genetics, University Hospital, Catholic University of Leuven, Leuven, Belgium
[3]Department of Family Medicine, Faculty of Medicine, Afyon Kocatepe University, Afyonkarahisar, Turkey

Corresponding author: Y. Soysal
E-mail: yasemin_soysal@yahoo.com

Genet. Mol. Res. 10 (3): 2148-2154 (2011)
Received February 21, 2011
Accepted August 16, 2011
Published September 21, 2011
DOI http://dx.doi.org/10.4238/vol10-3gmr1339

ABSTRACT. We present a 12-year-old girl with karyotype 46,XX. A comparative genomic hybridization array revealed a 3.172-Mb microduplication on 22q11.2. This chromosome 22q11.2 region microduplication has been described in patients with variable phenotypes; a large majority of them have identical 3-Mb duplications. The girl presented mild mental motor retardation, facial dysmorphism consisting of a long narrow face, widely spaced eyes, downslanting palpebral fissures, broad nasal base, short philtrum, thin upper lip, micro/retrognathia, low set and retroverted ears, microcephaly, high-arched palate, hypoplastic teeth, and hypernasal speech. She had delayed psychomotor development and behavioral problems. Molecular characterization of patients differs greatly among reports and detailed molecular characterization and documentation are needed to better understand the effects of these duplications. This description of the phenotype of a patient with microduplication on 22q11.2 will contribute

to the growing knowledge regarding deletions and duplications of the 22q11.2 region; this is important to conclude whether 22q11.2 duplication is a microduplication syndrome or not.

Key words: Array-CGH; Hypernasal speech; Temporal hair loss; Microduplication 22q11.2; Mental retardation

