Taiwanese Journal of Obstetrics & Gynecology 55 (2016) 288–292

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com

Case Report

Prenatal diagnosis and molecular cytogenetic characterization of *de novo* partial monosomy 3p (3p26.3→pter) and partial trisomy 16q (16q23.1→qter)

Chih-Ping Chen [a, b, c, d, e, f, *], Fung-Yu Hung [g], Schu-Rern Chern [b], Peih-Shan Wu [h], Yen-Ni Chen [a], Shin-Wen Chen [a], Chen-Chi Lee [a], Wayseen Wang [b, i]

[a] *Department of Obstetrics and Gynecology, MacKay Memorial Hospital, Taipei, Taiwan*
[b] *Department of Medical Research, MacKay Memorial Hospital, Taipei, Taiwan*
[c] *Department of Biotechnology, Asia University, Taichung, Taiwan*
[d] *School of Chinese Medicine, College of Chinese Medicine, China Medical University, Taichung, Taiwan*
[e] *Institute of Clinical and Community Health Nursing, National Yang-Ming University, Taipei, Taiwan*
[f] *Department of Obstetrics and Gynecology, School of Medicine, National Yang-Ming University, Taipei, Taiwan*
[g] *Department of Obstetrics and Gynecology, Hsinchu MacKay Memorial Hospital, Hsinchu, Taiwan*
[h] *Gene Biodesign Company, Ltd, Taipei, Taiwan*
[i] *Department of Bioengineering, Tatung University, Taipei, Taiwan*

ARTICLE INFO

Article history:
Accepted 24 February 2016

Keywords:
array comparative genomic hybridization
3p deletion
16q duplication
prenatal diagnosis
quantitative fluorescent polymerase chain reaction

ABSTRACT

Objective: To present the prenatal diagnosis and molecular cytogenetic characterization of a *de novo* unbalanced reciprocal translocation.

Case Report: A 37-year-old woman, G3P1, underwent amniocentesis at 17 weeks of gestation because of her advanced maternal age. Her husband was 38 years old. Amniocentesis revealed a derivative chromosome 3 with the deletion of terminal 3p and the addendum of an unknown extra chromosomal segment on the distal 3p. The parental karyotypes were normal. Prenatal ultrasound findings were unremarkable. Array comparative genomic hybridization (aCGH) analysis using cultured amniocytes revealed a 2.38-Mb deletion in 3p26.3 [arr 3p26.3 (1-2,380,760)×1] encompassing 15 genes, which included 3 OMIM genes *CHL1*, *CNTN6*, and *CNTN4*, and a 13.17-Mb duplication in 16q23.1-q24.3 [arr 16q23.1q24.3 (76,999,082-90,170,596)×3] encompassing 207 genes, which included 81 OMIM genes. The pregnancy was subsequently terminated, and a malformed fetus was delivered with facial dysmorphism. Postnatal cord blood analysis revealed a karyotype of 46,XY,der(3)t(3;16)(p26.3;q23.1)dn. Polymorphic DNA marker analysis by quantitative fluorescent polymerase chain reaction (QF-PCR) on the DNAs extracted from the placenta and parental blood showed a paternal origin of the aberrant chromosome.

Conclusion: The aCGH and QF-PCR analyses helped in delineating the genomic imbalance and parental origin of prenatally detected *de novo* unbalanced reciprocal translocation.

Introduction

Concomitant partial monosomy 3p and partial trisomy 16q is very unusual. To our knowledge, only one case has been previously reported [1]. Dikmetas et al [1] reported an infant with the karyotype of 46,XY,der(3)t(3;16)(p25;q13)mat associated with partial trisomy 16q (16q13→qter) and partial monosomy 3p (3p25→pter), anterior segment dysgenesis with iris hypoplasia on the right and glaucoma on the left, facial dysmorphism of synophrys, high prominent forehead, bitemporal narrowing, depressed nasal bridge, long philtrum and lingual phrenulum, atrial septal defect, patent ductus arteriosus, buphthalmos, stromal edema of the left eyes, right-sided atypical iris coloboma, and mild delay in growth and mental development.

In this paper, we also report the prenatal diagnosis and molecular cytogenetic characterization of a fetus with the karyotype of

* Corresponding author. Department of Obstetrics and Gynecology, Mackay Memorial Hospital, 92, Section 2, Chung-Shan North Road, Taipei 10449, Taiwan.
E-mail address: cpc_mmh@yahoo.com (C.-P. Chen).

http://dx.doi.org/10.1016/j.tjog.2016.02.015

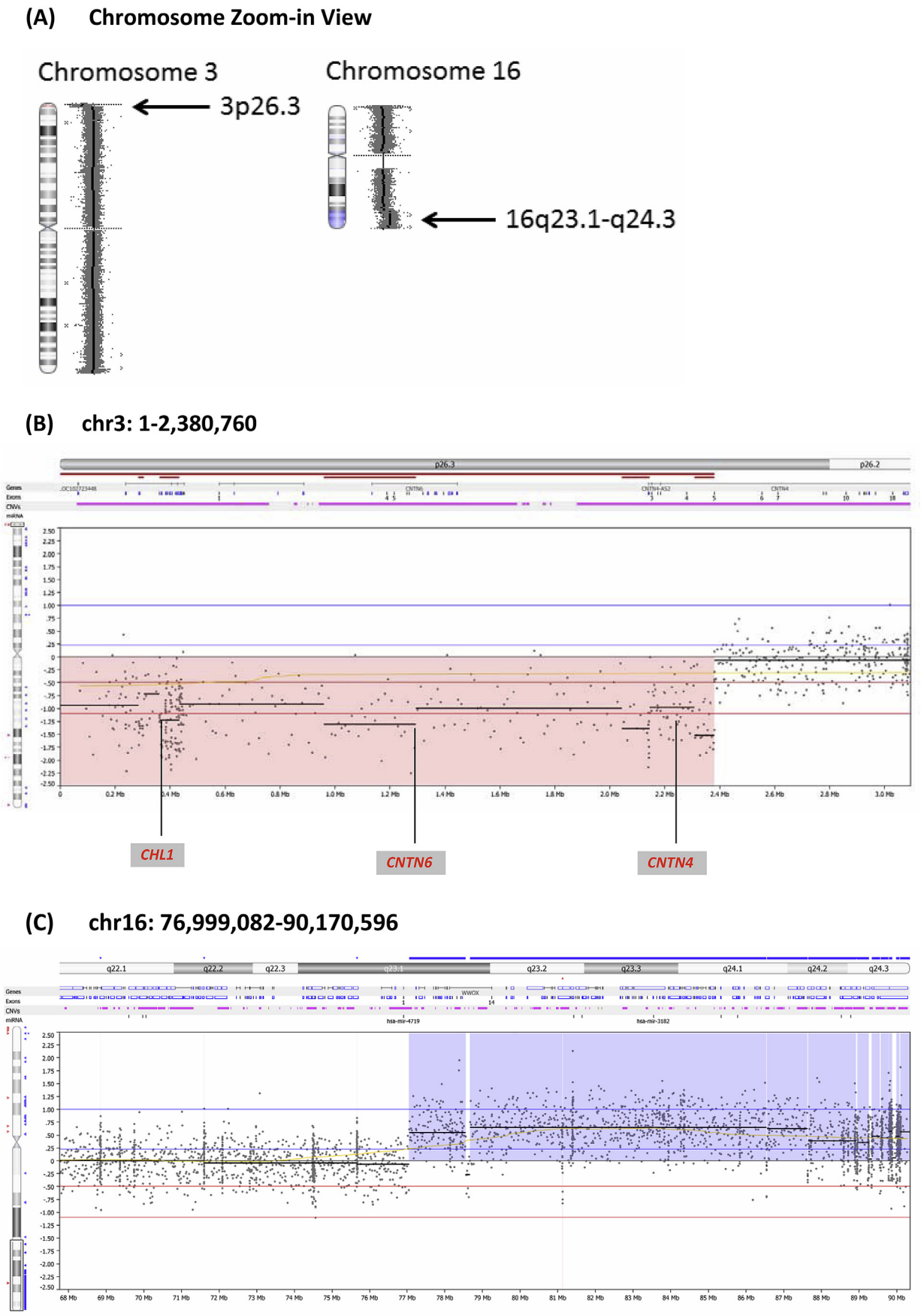

Figure 1. Array comparative genomic hybridization of uncultured amniocytes shows a 2.38-Mb deletion in 3p26.3 [arr 3p26.3 (1-2,380,760)×1] and a 13.17-Mb duplication in 16q23.1-q24.3 [arr 16q23.1q24.3 (76,999,082-90,170,596)×3]. (A) Chromosome zoom-in view. (B) Chromosome 3. (C) Chromosome 16.