A Deletion and a Duplication in the same Chromosome by Array CGH

C. Garrido[1], D. Escué[1], M.A. Caamaño[1], A. Morales[1], M. Naharro[1], D. Visus[1], S. Sánchez[1], D. Trujillano[1], N. Guerrero[2], P Carrasco[3], E. Geán[1]

[1]**Reference Laboratory** (BCN, Spain)
[2]**Hospital Juan Ramón Jiménez,** Neuropediatrics Unit (Huelva, Spain)
[3]**Hospital Juan Ramón Jiménez**, Genetics Unit (Huelva, Spain)

About the Author

Carles Garrido specialised in molecular genetics after graduating from his MSc in Molecular Biology in 2008 at The Autonomous University of Barcelona. Within the field of molecular genetics, Carles is an expert in FISH and cytogenetics. He has worked in the department of genetics and prenatal genetics at Hospital San Juan de Dios in Barcelona and for the last 9 years has been head of the department FISH and Arrays at Reference Laboratory, where he has carried out the analysis of over 3,000 samples using array CGH. Throughout his career he has written various national and international publications related to the different areas in which he has worked.

Introduction

Partial duplications and deletions in the same chromosome are very rare events, usually sporadic. They could be due to a parental paracentric inversion of that chromosome, non-allelic homologous recombination (NAHR) or to an abnormal cross-linking during meiosis. Also, the phenotypic expression of both aneuploidies could be highly variable and the etiological mechanism is complex.

Case report

- **The patient, a 6-years-old girl,** was referred to us because she had growth retardation and traits consistent with autistic spectrum disorder. The mother had developed a normal pregnancy, with delivery at 32 week of gestation.
- **Good health generally,** the face with typical characteristics of prematurity showing trigonocephaly and bilateral exophthalmus. Short stature. Lack of eye contact and no relationship with environment. Normal cranial nerves. Slightly increased tone in lower limbs with symmetrical reflexes.
- **Autistics traits:** poor language, lack of comprehension, does not respond when others interact with her and she does not point. Adequate cerebral growth rate.
- **Normal cytogenetic study** was conducted two years ago. We performed array CGH in peripheral blood. Cytogenetic studies were carried out in the parents after the child's results.

Results

- The patient shows a deletion and a contiguous duplication in chromosome 7, at the region adjacent to the ELN gene without affecting it:

arr[GRCh37] 7q11.21q11.23(62030284_72401145)x1, 7q11.23q21.11(75061927_83488169)x3

- Cytogenetic studies in the parents showed a paracentric inversion in the long arm of chromosome 7: 46,XX,inv(7)(q11.21q21.1) in the mother, whilst in the father no structural or numerical chromosome abnormalities were observed.

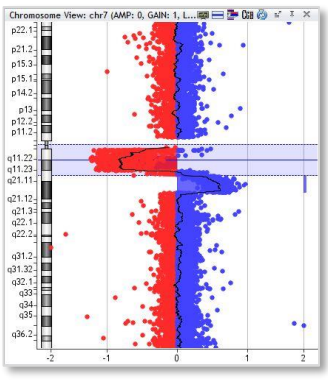

Figure 1. arr[GRCh37] 7q11.21q11.23(62030284_72401145)x1,7q11.23q21.11(75061927_83488169)x3

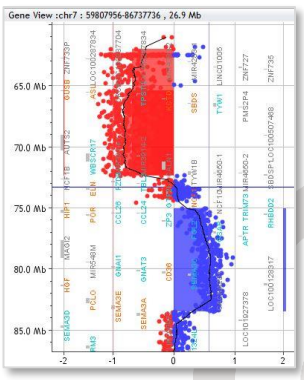

Figure 2. 7q deletion and 7q duplication in detail, ELN gene is normal.

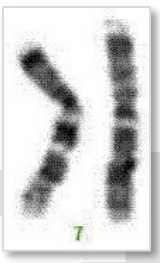

Figure 3. Girl karyotype: 46, XX, With 400 G-bands.

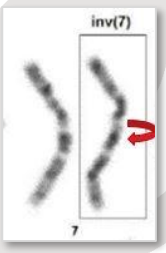

Figure 4. Mother karyotype: 46,XX,inv(7)(q11.21q21.1), with 400 G-bands

Discussion

Array CGH has allowed identification of the aetiology of the patient: 7q11.21q11.23 interstitial deletion and 7q11.23q21.11 interstitial amplification. The existence of paracentric inversion in the mother completely changed the risk of recurrence for the parents in case of future pregnancies, getting personalized genetic counselling.

As far as we know this is the only case in literature, with a deletion contiguous to duplication of this region in chromosome 7, with normal dose of ELN gene.

The authors certify that they have no conflict of interest.

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