Characterization of a rare analphoid sSMC(7)

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sSMC

47,XY,+mar[10]/46,XY[40]
Mosaic 20%

Mosaic 73%

46,XX

47,XY,+mar/46,XY

46,XY

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BAC Probes

α-Satellite probes

WCP Probes

BAC Probes
• Chr 15
• Chr 13/21
• Chr 14/22
• Chr X and Y
• ..... 
• ....
• Chr 7
• ..... 
• .....
What looks like this sSMC?
BAC Probes

RP11-237G17 (7q34) —

RP11-298A10 (7q35) +

TelVysion (7qter) ++

47,XY,+mar[10]/46,XY[40].ish invdup(7)(qter→q35::q35→neo→qter)(wcp7+,D7Z1-,RP11-298A10+,TelVysion 7q++)

Permanent Working Group Marker Chromosomes_ ECC 2015
47,XY,+mar[10]/46,XY[40].ish invdup(7)(qter→q35::q35→neo→qter)(wcp7+,D7Z1-,RP11-298A10+,TelVysion 7q++).

arr[hg19] 7q35q36(143696249-159119707)x2~3
Fenotype/genotype correlations
INTRODUCTION

Aneuploid small supernumerary marker chromosomes (SSCMCs) are a rare subclass of Chromosomally Stable SAC that are regarded as alpha-satellite DNA. These marker chromosomes cannot be identified accurately by conventional banding techniques alone (being necessary to apply molecular cytogenetic methods in favor of a detailed characterization [1]).

Approximately 3-5% of abortions had been described, with this involving 20 of the 22 autosomes and both sex chromosomes, with about 10% resulting from chromosomes 15, 13 and 8 [2]. Only three cases are in SSCMCs [2], but few have been reported with a specific allele [1].

In terms of clinical findings, there are three major groups to be considered: no clinical consequences (50%), moderate to severe clinical consequences (30%) and the largest group, the most severe clinical consequences. If an aneuploid SSCMC does not cause aneuploidy, the presence of molecular clinical findings may be absent, or in case of a rare chromosomal aneuploidy, the patient may present a mosaic clinical picture [3].

We report a child with several dysmorphic features and severe developmental delay presenting a de novo aneuploidy SSCMC in mosaic. This was detected through fluorescence in situ hybridization performed on the terminal long arm of chromosome 2, using a breakpoint concerning a part of chromosome 2.

RESULTS & DISCUSSION

Figure 1 - (A) karyotype showing a balanced translocation (46,XY,t(2;12) (q32.3;q23.1)) identifying the patient’s parents. (B) CGH analysis showing the presence of microdeletions in the long arm of chromosome 2, including the terminal region of chromosome 2. (C) FISH analysis showing the presence of microdeletions of the long arm of chromosome 2, including the terminal region of chromosome 2. (D) FISH analysis showing the presence of microdeletions of the long arm of chromosome 2, including the terminal region of chromosome 2.

Figure 2 - (A) Microarray analysis showing the microdeletions in the long arm of chromosome 2. (B) Chromosome 2 showing the deletion region in question. (C) Genes associated to the deleted region in question (D). (D) Genes associated to the deleted region in question (E).

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REFERENCES

1. Bárbara Marques, Filomena Brito, Cristina Alves, Sónia Pedro, Cristina Ferreira, Marta Amorim, Hildeberto Correia. 1
4. Permanent Working Group_ Marker Chromosomes_ ECC 2015
Thanks for your attention!!!