XLIII Conferências de Genética
Doutor Jacinto Magalhães

28 MARÇO DE 2014

"A GENÉTICA DO X"

CGM
Centro de Genética Médica
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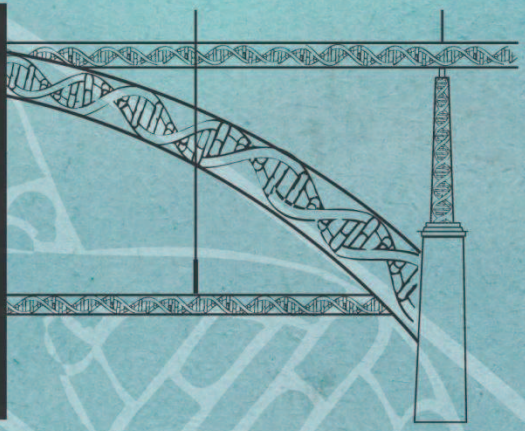

Cromossomopatias
Doenças Hereditárias do Metabolismo
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Genómica nas Doenças Neuromusculares
Aconselhamento Genético e Ética

Submissão de resumos até 9 de Março de 2014

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STUDY OF THE FMR1 GENE STRUCTURE AMONG WOMEN WITH OVARIAN DYSFUNCTION FROM THE BASQUE COUNTRY

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FMR1 premutation and intermediate alleles have been associated with the development of different forms of ovarian dysfunction, being the Premature Ovarian Failure (POF) the most serious one. A group of 68 women with ovarian dysfunction of unknown aetiology and 47 control women from the Basque Country has been analyzed. Considering the number of CGG repeats, the frequency of alleles with ≥35 CGG repeats was statistically higher in the whole patient group (12.50% vs. 0%). Concerning their ovarian condition, the patient group was divided in three categories and, in the three subgroups the alleles with ≥35 CGG were also statistically higher than in controls. As the AGG interspersion pattern seems to be correlated with the instability of the alleles, the CGG repeat internal structures have been analyzed. Many of the intermediate and premutation alleles found in the patient group appeared to have two interruptions with more than 15 CGG at the 3' end (65%). Interestingly, among these alleles the predominant structure was 9+9+n, indicating a loss of AGG interruptions at the 3' end. Therefore, the data showed that among patients the alleles were more unstable and that this instability influencing the FMR1 expansion might be related with the development of an ovarian dysfunction.