

# Oral communication

## Comunicação oral

### CO-01

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

Maitane Barasoain<sup>1</sup>, Gorka Barrenetxea<sup>2,3</sup>, Iratxe Huerta<sup>1,4</sup>, Mercedes Téllez<sup>1,4</sup>, Amaia Carrillo<sup>2</sup>, Cristina Pérez<sup>2</sup>, Eduardo Ortiz-Lastra<sup>3</sup>, Javier González<sup>3</sup>, Begoña Criado<sup>5</sup>, Isabel Arrieta<sup>1</sup>

<sup>1</sup> Department of Genetics, Physical Anthropology and Animal physiology, Faculty of Science and Technology, University of the Basque Country, Bilbao, Spain;

<sup>2</sup> Center for Reproductive Medicine and Infertility Quirón Bilbao, Bilbao, Spain;

<sup>3</sup> Department of Medical-Surgical Specialities, Faculty of Medicine, University of the Basque Country, Bilbao, Spain;

<sup>4</sup> Virgen de Begoña Clinical Analysis Laboratory (Medikosta), Erandio, Spain;

<sup>5</sup> Cooperativa de Ensino Superior Politécnico e Universitário (CESPU), Porto, Portugal

maitane.barasoain@ehu.es

*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  $\geq 35$  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  $\geq 35$  CGG were also statistically higher than in controls. As the AGG interspersed 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.