

## Abstract 91

### EFFECT OF AN AMINO ACID POOL ON FRAGILE X SYNDROME CARRIERS: CLINICAL AND NEUROCHEMICAL PRECLINICAL STUDY

**Ingrid Barbato<sup>1</sup>, Tania de Liz<sup>1,2</sup>, Luismar Porto<sup>2</sup>, Carla Tasca<sup>2</sup>**

<sup>1</sup>Neurogene Laboratory, Florianópolis, Santa Catarina, Brazil

<sup>2</sup>Federal University of Santa Catarina Florianópolis, Santa Catarina, Brazil

This work reports on therapeutic effects observed in Fragile X Syndrome (FXS) patients after the intake of a pool of amino acids known as Face®. To further evaluate clinical observations, a randomized, double blind clinical study, and the application of psychometric tests were performed. Pre-clinical animal studies were also considered. In particular, neurochemical experiments were carried out in rats (*Rattus norvegicus*). The clinical evaluation was performed by analyzing a questionnaire (before and after intervention) applied to 43 FXS patients. Significant improvement has been observed in symptoms such as learning, behavior and emotion of affected and gray zone individuals. The following psychometric exams were applied (before and after intervention) to 13 affected individuals: Autistic traits evaluation (ATA Test), Mini-Exam of Mental State, and Vineland Adaptive Behavior Scales. The results for the three tests were significant for the affected individuals, showing important positive changes in cognitive traits, learning, social behavior and, surprisingly, with respect to their autistic characteristics. For the laboratory experiments, 24 rats were orally treated with Face® and saline solution as control during 60 days, and their hippocampus, cerebellum and cortex were analyzed. FMRP and mGluR1 were quantitatively determined in these brain structures, as well as the activity of glutamate transporters, ammonia concentration and cell viability. Significant changes were found in the Na<sup>+</sup>-dependent and Na<sup>+</sup>-independent glutamate transporters in hippocampus, after 30 days of intake (p<0.001). After 45 days an increase of 49.9% in the level of FRMP was observed (p=0.048). In the hippocampus the amount of mGluR1 decreased by 48.3%, while in the cortex, after 45 days it decreased by 18.9% in rats treated with Face®. Face® has shown to be a nontoxic product, which activates the glutamatergic system. It produced significant improvement on mental and autistic patterns, social behavior, learning abilities and on general emotional states of the investigated patients. These results strongly suggest a positive effect of the amino acid pool on the FXS phenotype.

Acknowledgements: We thank Mr. José Guerra from LBE Laboratory (São José-SC, Brazil) for kindly providing us with the amino acid pool Face® for this research.