**P-003 - SCREENING FOR 9 COMMON JEWISH GENETIC DISEASES IN 441 ASHKENAZI JEWS IN ARGENTINA**

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***INTRODUCTION:*** One in five individuals of Ashkenazi Jewish descent carry a mutation for a recessive disease. There is little information available about the frequency and prevalence of inherited disorders in this population in Argentina, despite being the sixth largest Jewish community in the world and the largest in Latin America. ***OBJECTIVES:*** Screening for 9 common Jewish genetic diseases in Argentinian Jews who have at least one Ashkenazi grandparent. The project aims at raising awareness about the high prevalence of Ashkenazi Jewish genetic mutations and diseases. ***MATERIALS AND METHODS:*** During 2018, Ierusha Foundation offered several educational lectures in Jewish organizations throughout Buenos Aires, where attendants could get a free non invasive genetic testing through buccal epithelial cells. The results were returned by a geneticist, who provides genetic counseling. A total of 441 samples were screened for the following 9 monogenic disorders: Fanconi Anemia (FANCC), Cystic Fibrosis (CFRT), Glycogen storage disease type 1 (G6PC), Maple Syrup Urine Disease (BCKDHB), Tay Sachs (HEXA), Canavan (ASPA), Gaucher (GBA), Hyperinsulinism (ABCC8), Niemann Pick (SMPD1). The samples were analyzed at Genia Molecular Genetics Laboratory, using the DNeasy® Blood & Tissue Kit from Qiagen® for extraction and purification of genomic DNA. The amplification was carried out through a pool of primers designed with AmpliSeq ™ technology and the sequencing of the amplified regions through Post-LightTM Ion Semicondutor Sequencing (Next Generation Sequencing). ***RESULTS:*** Among the 441 samples screened, 138 mutations were found (31,29%): CFTR: 69 cases (50%), GBA: 40 (28,98%), ASPA: 11 (7,97%), G6PC: 10 (7,24%), HEXA: 12 (8,69 %), ABCC8: 6 (4,34%), BCKDHB: 5 (3,62%), SMPD1: 3 (2,17%), FANCC: 1 (0,72%). 21 cases were double heterozygous and 5 cases homozygous: 4 CFTR, 1 GBA ***CONCLUSIONS:*** The high rate of mutations detected (31,29%) represents an incidence of 1 in 3, higher than expected in this population. This new data about the Argentinian Jewish Ashkenazi community will contribute to the prevention of genetic diseases and provide new epidemiological information.