Genomics Of Signalopathies At The Service Of Medicine, Medical University of Sfax, Sfax, Tunisia
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Introduction: Wilson disease (WD) is an autosomal recessive genetic disorder caused by loss-of-function mutations in the P-type copper ATPase, ATP7B (ATPase copper-transporting beta), which transports copper out of cells. It is characterized by toxic accumulation of copper primarily in the liver and brain, leading to liver disorders and/or neuropsychiatric symptoms.
Objectives: Here, we report a Tunisian pedigree associated to familial ATP7B gene mutation.
Methods: Medical genetic investigations, and molecular screening of ATP7B gene mutations were performed to a Tunisian three-generation pedigree with eight members having neuropsychiatric symptoms. Molecular genetic testing of the ATP7B 21 exons was carried out by direct sequencing.
Results: A compound heterozygote mutational status of ATP7B with 2 substitutions: p.H1069Q and p.D642H was found. The family originated from the city of Sfax (Tunisia) showed a pronounced amount of consanguinity and eight members affected by WD. All cases derived from consanguineous couples and harbored psychiatric disorders associated or not to neurologic symptoms. Diagnosis of WD was piloted first through the cases harbouring intention tremor in the upper limbs and ataxia associated with psychiatric symptoms.
Conclusions: The first missense mutation p.H1069Q - c.3207C>A (CAC-CAA) (exon 14) is the most commonest mutation in WD associated with late onset neurological conditions in Europe (Natural variantiVAR_000758 dbSNP:rs76151636). The second missense mutation in exon 6 : p.D642H - c.1924G>C (GAC-CAC) (Natural variantiVAR_000713 dbSNP:rs72552285) affects the domain affinity to copper or the folding structure in the cytoplasmic region and decreases the stability, leading to abnormal localization of the protein within cytoplasm and an impairment of protein function.

Disclosure: No significant relationships.
Keywords: ATP7B; Copper; Neuro-psychiatric symptoms; Wilson disease

EPV0333

A populational review of the amyloid precursor protein gene mutations relevant to alzheimer's disease

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Introduction: The genetic component of Alzheimer's disease was previously studied and more than sixty amyloid precursor protein (APP) gene mutations were identified. However, the populational aspects of this component were scarcely discussed despite that many of the reports mentioned the demographic ancestry of the carriers or probands.
Objectives: In this short study, we aimed to review the APP gene mutations relevant to Alzheimer's disease from a Populational Genetics point of view by evaluating the current literature for the demographic description of the carriers or families in which the mutations were identified.
Methods: In this regard, multiple genetic studies on the APP gene mutations relevant to Alzheimer's disease were reviewed and the incidence of the mutations was analyzed considering the ancestry of the patients.
Results: We found many possible scenarios regarding the incidence of the APP gene mutations in Alzheimer's disease patients and general population. On the one hand, we could identify several mutations which were present in more than one population (eg. V615M, V717I, V717L) and on the other hand, some mutations could be observed in certain populations (eg. E693delta, the Osaka mutation, which was until now observed in Japanese patients, while E693G was found in a Swedish family). One particular case is that of the isolated populations (eg. the Icelandic population in which an APP mutation protecting against Alzheimer's disease is more frequent in the general population as compared to the patients).
Conclusions: We were able to identify several mutations which were characteristic to many populations, but also some population-specific features regarding the APP genotypes.

Disclosure: No significant relationships.
Keywords: Alzheimer's disease; amyloid precursor protein; Populational Genetics

EPV0334

Association between IL-17, IL-23 with neurocognitive scales in patients with Alzheimer's disease

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Introduction: Alzheimer's disease (AD) is a degenerative brain disease and the most common cause of dementia. Evidence suggests that various cytokines, including interleukins (IL) IL-6, IL-10, IL-12 are actively involved in the pathogenesis of AD. The role of IL-17 and IL-23 is less clear.
Objectives: To investigate the correlations between IL-17, IL-23, and neurocognitive scales in patients with Alzheimer's disease.
Methods: The study included 45 patients: 15 patients with Alzheimer's disease and 30 patients without cognitive deficit (control group). Clinical and psychometrical methods were used: Mini Mental State Examination (MMSE) scale; Montreal Cognitive Assessment (MoCA), Frontal Assessment Battery (FAB), Alzheimer Disease Assessment Scale-cognitive (ADAS– cog). Serum levels of cytokines of IL-17 and IL-23 were analyzed by sandwich ELISA on "Chem Well 2900" immunoanalyzer (Awareness Technology, USA).
Results: A significantly positive correlation was observed between IL-17 and IL-23 for all AD patients ($r = 0.723$, $p = 0.002$). A significant inverse correlation was observed between serum concentration of IL-17 and MoCA score ($r = -1.0$, $p \leq .0001$) and IL-23 and MMSE score ($r = -0.553$, $p = 0.032$) in all AD patients. However, no other significant correlations were found between IL-17 and the

scores MMSE, FAB, ADAS- cog and between IL-23 and the scores MoCA, FAB and ADAS- cog.
Conclusions: Proinflammatory cytokines (such as IL-17 and IL-23) have been associated with cognitive impairment. However, the complicated relationships of the two cytokines with the pathogenesis of AD need to be further investigated in the future.

Disclosure: No significant relationships.
Keywords: Alzheimer's disease; inflammation; interleukin; neurocognitive scales.

EPV0335

The impact of the environment and parental education on the emotional state of adolescents

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Introduction: Its basic object of study is the understanding of the individual and the group, Scientific direction in the study of behavior is an important feature of psychiatry.
Objectives: Adolescence is a stage of physical and mental development of human, usually listed between the stage of childhood and legal maturity. in the middle aged 13 and 18 years the sexual way begins.
Methods: Education is the primary category of pedagogical theory, which includes the concepts of education and teaching. Physical education is a subject taught in school to train students physically, to enable them to work and to defend themselves. The totality of spiritual, mental and physical qualities or qualities of family and society and learning and working.
Results: By taking preventive measures to close educational institutions against the spread of the covid virus 19, it has affected the education system of students, teachers and parents because it has been physically disconnected from learning and has gone virtually ONLINE, where it has been a form of home isolation.
Conclusions: Parenting is a constant concern for our entire society today, for child and adolescent psychiatry specialists and education experts, especially for all young parents.

Disclosure: No significant relationships.
Keyword: beli

EPV0336

Protective role of lipoic acid in the prevention of oxidative stress caused by cadmium in the vascular endothelium of rats

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Introduction: Acute and chronic exposure to cadmium (Cd), due to its increased use and application in the industry, can result in the development of atherosclerosis, the occurrence of cardiomyopathy, cerebrovascular stroke, as well as carcinogenesis in some organs. The target for cadmium action is thought to be the vascular endothelium.
Objectives: The aim of this study is to investigate the occurrence of oxidative stress on blood vessel endothelium induced by subacute administration of cadmium, as well as the protective power of alpha-lipoic acid (α-LA) supplementation on the Wistar strain albino rat model system.
Methods: After anesthesia of rats in the vivarium of the Scientific Research Center for Biomedicine, Faculty of Medicine in Niš, blood was collected by cardiac puncture and sent to the Clinic of Nephrology, Clinical Center in Niš, Serbia for determination of hematological parameters.
Results: According to the results of this study, it can be seen that the number of granulocytes is reduced due to cadmium intoxication, which is probably induced by the migration of neutrophils into tissues. The number of lymphocytes was increased due to subacute cadmium intoxication compared to the control group of animals. The positive efficacy of α-LA supplements in combating the adverse effects of cadmium on blood vessels is also confirmed.
Conclusions: Cadmium administration is thought to cause a systemic inflammatory reaction due to the formation of free radicals in the blood vessel endothelium. Administration of α-LA supplement confirms that it can be used as an antioxidant in the clinical management of many diseases and also in cadmium intoxication.

Disclosure: No significant relationships.
Keywords: cadmium; blood vessel; antioxidant; oxidative stress; alpha-lipoic acid

EPV0337

Glutathione as a powerful antioxidant in oxidative stress in the brain tissue of rats caused by the pathophysiological action of copper

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Introduction: Due to increased human activity, the amount of copper in air, soil and water has increased. Copper, at minimum concentrations, is essential for the normal functioning of the organism (cellular respiration, hemoglobin formation, growth and reproduction). At higher concentrations, copper is deposited in the liver, brain tissue, and bone marrow.
Objectives: To investigate the protective role of the supplement, glutathione (GSH), the S-donor ligand, in conditions of chronic copper intoxication via the parameters of oxidative stress, ie. Alkaline and acidic DNase values in brain tissue in albino rats of Wistar strain.
Methods: The model system for testing the effects of copper exposure and the protective effect of GSH was a study on female albino rats of Wistar strain, stored in the vivarium of the Scientific Research Center for Biomedicine, Faculty of Medicine, Niš, Serbia.