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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

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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