E-P164: Impact of Happiness Educational and Performance Program of Fordyce on Depression Severity among Patients Undergoing Hemodialysis in Urmia city hospitals, Iran: A Randomized Controlled Trial

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Background: Depression has a high prevalence in hemodialysis patients. Fordyce's happiness training and performance as a complementary therapy may improve mental status and reduce complications, including depression. This study was conducted with the aim of investigating the effect of this program on the severity of depression of hemodialysis patients in Urmia hospitals .

Materials and methods: In this clinical trial, 60 hemodialysis patients were randomly divided into two control groups (30 people) and an intervention (30 people). The Fordyce program was conducted in each hemodialysis session for 30 minutes and six sessions for the experimental group through communication, interviewing, and listening to solve their problems. Only the experimental group was considered. Two demographic and depression questionnaires were used to collect data, which were analyzed using SPSS software (Code of ethics: IR.UMSU.REC.1400.368) .

Results: In comparison with the experimental group, a significant difference was shown in the mean depression scores in the experimental group immediately after the intervention and four weeks after the intervention ($p<0.05$).

Conclusion: The findings of the present study showed that this program significantly reduces depression scores in hemodialysis patients. Further research with larger sample size is expected to confirm the results obtained and integrate the Fordyce Happiness Program into clinical practice.

Keywords: happiness, program, depression, hemodialysis

E-P165: The first report of Charcot-Marie-Tooth disease, type 4G (CMT4G) in Iran

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Background: Charcot-Marie-Tooth (CMT) is the most common type of inherited peripheral neuropathy and its autosomal recessive forms (ARCMT) account for less than 10% of the patients in Europe but are more frequent (30–50%) in populations with high rates of consanguinity. CMT type 4G, also named hereditary motor and sensory neuropathy-Russe (HMSN-R) was first reported in Balkan Gypsies as a myelinopathy starting with a progressive distal lower limb weakness, followed by upper limb involvement and prominent distal sensory impairment later in the patient's life .

So far, only one homozygous variant (g.9712G>C) in the 5'-UTR of specific HK1 gene isoforms has been associated with HMSN-R, described as a founder mutation in the gypsy population. HK1 gene encodes hexokinase 1 protein, and its 5` untranslated exons undergo extensive alternative splicing generating different isoforms, some of which are tissuespecific. This mutation in HK1 is now considered one of the most common causes of ARCMT in European Roma communities. Given the prevalence of consanguineous marriage in Iran, ARCMT is expected to be more prevalent, and CMT-4D, CMT-4C, and CMT-4A patients are so far reported in this population .

Method and Materials: The phenotype of a consanguineous Iranian family was characterized and whole-exome sequencing was performed to identify the disease-causing variant in this family. The generated Fats files were analyzed using the GATK pipeline, adhering to best practices. Furthermore, the GATK germline CNV detection pipeline was used to detect copy number variations (CNVs) .

Results: We present a 21-year-old female manifesting progressive muscle weakness in distal limbs since 11-year-old and diagnosed with chronic demyelinating sensorimotor polyneuropathy. A homozygous variant defined as c.19C>T (p.Arg7X) in HK1 gene (NM_033498) was detected in this individual, which co-segregates in the family and is predicted to cause loss of normal protein function either through protein truncation or nonsense-mediated mRNA decay. This variant is located in the critical HMSN-Russe region present in specific HK1 isoforms similar to the g.9712G>C variant. Therefore, we speculate similar consequences, leading to the CMT4G phenotype in this patient.

Conclusion: To our knowledge, this is the first observation of CMT4G in the Iranian population. The same Arg7X variant is recently observed in another unrelated CMT patient from Pakistan, proposing a possible prevalence of this variant in the Middle Eastern populations. In conclusion, we expand the molecular spectrum of the HK1-related CMT disorder and provide supporting evidence for the observation of CMT4G outside the Roman population.

Keywords: CMT4G, Whole-exome Sequencing, HK1