

**Aim:** We aimed to carry out a linkage analysis in 6 Iranian families to find an association between the FEB1 and GEFS+.

**Introduction:** Generalized Epilepsy with Febrile Seizures plus (GEFS+), is a group of genetic epilepsy syndromes, likely to commence in the first year of life, in which, patient presents with febrile and tonic-clonic seizures. GEFS+ is associated with an autosomal dominant pattern and is caused by mutations in SCN1B which encodes the beta 1 subunit of sodium channels.

**Methods:** We conducted a case-control study in January 2017, with 6 families, with a total of 35 members entering the study with convenience sampling method, within which, 12 members were as the case group, diagnosed with autosomal dominant GEFS+, hospitalized in Ali Asghar Children's hospital, Iran University of Medical Sciences. 23 family members with no diagnosed GEFS+ were as the control group. Written consent was obtained from all family members according to the protocols of the ethics committee of the university. Afterwards, using D8S533 marker for FEB1 gene, with a Logarithms of Odds (LOD) of 3.16, two-point linkage analysis and haplotype reconstruction was carried out using MLINK program and Simwalk2 respectively.

**Results:** Haplotype reconstruction analysis in the case group revealed a haplotype associated with GEFS+. However, in the control group, not such an haplotype was seen and the difference between two groups was significant ( $p < 0.05$ ).

**Conclusion:** In this study we reported a strong linkage between GEFS+ and FEB1 gene. This may clarify the etiology underlying GEFS+ and gives us chances in GEFS+ screening using FEB1.

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### PS037

#### Genetical variability of VP1 gene of BK virus in HIV-infected patients



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**Aim:** The aims of this study were: to determine the prevalence of BK viruria in HIV-infected patients, to determine the distribution of BKV subtypes and the presence of nucleotide substitutions and mutations in the VP1 gene of BKV isolates.

**Introduction:** A broad range of diseases associated with BK virus (BKV) such as nephritis, haemorrhagic cystitis, encephalitis, retinitis and pneumonia have been reported in HIV-infected patients over the last few years. However, these diseases do not occur in all HIV-infected patients, suggesting that other factors, such as genetic variability of BKV, can contribute to their occurrence. Mutations in the BC loop of the VP1 gene may lead to selection of more aggressive variants of BKV.

**Methods:** The study included 50 HIV-infected patients. Sem-nested PCR was used for amplification of 290-nt fragment within the VP1 gene and all the positive PCR products were then directly sequenced. The sequence analysis was performed by using the appropriate bioinformatics tools.

**Results:** The frequency of BK viruria in HIV-infected patients was 56%. The predominant BKV subtype was I, followed by subtype IV. The majority of mutations were located within BC loop of VP1. The most frequent mutation was E82D.

**Conclusion:** The increased levels of BKV replication are associated with a higher incidence of mutations in the BC loop of VP1, and mutations in this domain may lead to changed tropism and the selection of more aggressive variants of BKV. Further studies are needed in order to select the patients with a higher risk of developing BKV associated-diseases.

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### PS182

#### Cellular interaction in central and peripheral immune organs due to chronic light stress



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**Aim:** Study cellular interaction in central and peripheral immune organs at prolonged all-day illumination in an experiment on rabbits.

**Introduction:** Prolonged all-day illumination is considered nowadays as one of the stress-factors for the living organism and causes malfunctions of the neuroendocrine system and may initial immune dysfunction.

**Methods:** Experimental rabbits ( $n = 10$ ) were in artificial lighting in the day and electric lighting at night during 12 months. Control animals ( $n = 5$ ) were kept in natural day and night lighting conditions. Cell density in immune organs (thymus, bone marrow, spleen) were measured in surface area which was determined by a rectangle  $100 \times 100 \mu\text{m}$ . The results were processed with standard statistical methods and reported as mean  $\pm$  standard deviation (SD).

**Results:** The cell density in the thymus and the bone marrow was decreased: in the cortex of the thymus was  $359.6 \pm 2.9$ , in the medullar part –  $250.8 \pm 2.9$ , in the bone marrow –  $176.4 \pm 2.9$  (cells in  $100 \times 100 \mu\text{m}$ ). An intensified formation of the connective tissue, an increasing of involutive processes and degenerative changes of lymphocytes were microscopically found in the spleen and the thymus. The cell density in the spleen was decreased too: in T zone –  $235.8 \pm 3.7$ , in B-zone –  $159.5 \pm 1.9$  (cells in  $100 \mu\text{m} \times 100 \mu\text{m}$ ). The causes of these changes, probably, may be decrease of the differentiation and migration of lymphocytes as result negative influence of the prolonged light on central immune organs.

**Conclusion:** These changes in organs of the immune system indicate both a premature aging of the spleen and the thymus and probably of all the immune system. Significant reduction in cell density in the immune organs associate with negative effects of the chronic light stress and leads to expressed immune dysfunction.

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### PS217

#### Intermittent low-level lead exposure causes anxiety and cardiorespiratory impairment



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