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CLINICAL ASPECTS OF SUSPECTED GENETIC EPILEPSY IN MULTIPLEX FAMILIES FROM REPUBLIC OF MOLDOVA - PRELIMINARY RESULTS FROM THE NEWLY CREATED NATIONAL EPILEPSY REGISTRY

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Until the advent of next-generation sequencing, due to phenotypic and genotypic heterogeneity and low-yield technologies for genetic testing - most epilepsy patients did not benefit from an accurate molecular diagnosis, even if they had pathologies with undeniable genetic aetiology. Research in human genetics has established that a genetic basis contributes to susceptibility to epilepsy in most cases. However, the multifactoriality of epilepsy, has made it a challenge to decipher its genetic architecture and to determine the specific genetic risks for each individual with epilepsy. To associate a gene with epilepsy for the first time, more cases without family ties or families with multiple affected members are needed. The aim of our study is carrying out a clinical-genetic study of multiplex families from the Republic of Moldova, for estimating the specific biomarkers and establishing their weight in epileptogenesis, for the elaboration of a family risk prediction score.

In order to achieve the set objective, an epidemiological, descriptive, cross-sectional study (2018-2023) was planned and already started with lancing a National Epilepsy Registry for multiplex families – families with at least 2 members (relatives of 1st degree) with definite epilepsy. Preliminary statistical methods were applied.

Our National Epilepsy Registry consists now of 68 families including 180 members, from which subjects with epilepsy (68), epilepsy relatives (32) and healthy relatives (80). The distribution by age of both groups, denotes a negative trend of cases of epilepsy with advancing age, with a peak in the 3rd decade of life. The distribution by sex groups denotes a numerical prevalence of female subjects in both studied subgroups, possibly due to the concept of "maternal effect", known in epilepsy. In our experimental group, the seizure onset, based on the available EEGs was mostly focal then generalized, but the supportive documentation was missing in a lot of cases. The seizure type was mostly represented by the motor variants, here included all the available kinds – tonic, tonic-clonic, clonic, myoclonic, motor automatisms etc. and concerning the awareness during the seizures, in the majority of the affected subjects - it was impaired.

We present the preliminary results of the epidemiological analysis of Moldavian multiplex families with epilepsy. Next step will be the trio genetic testing via Whole Exome Sequencing. The register is constantly being updated.