DNA detection on Guthrie-card resulted positive and a novel gene mutation in PLP1 was found at whole-exome-sequencing.

In both cases neurological impairment differential diagnosis between cCMV and genetic syndrome appeared particularly challenging. bMRI suggested for both a cCMV causative role; in case-1 radiologic imaging revision revealed a possible fetal clastic damage, in case-2 grey matter degeneration with normal white matter appeared inconsistent with PLP1 mutation. Moreover, a detailed perinatal history revealed early signs of cCMV, which were ignored: in case-1 intrauterine-growth-restriction, in case-2 pathological audiological screening.

In conclusion, congenital cytomegalovirus infection should always be considered in cases of infant with unexplained neurological impairment, particularly when signs suggestive of congenital infection are present at birth. In fact, an early screening of possible neurological sequelae allows precocious treatment whose efficacy is documented only if started within the first month of life.

### 386 EPILEPSY IN PEDIATRIC PATIENTS – EVALUATION OF ANATOMIC STRUCTURES' VOLUME OF THE BRAIN

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Epilepsy is a disease of the central nervous system with somatic, vegetative and psychological symptoms that appear as a result of various morphological and metabolic changes in the brain. Epileptic seizures are the manifestation of temporary problems with communication between nerve cells. About 1% of the world’s population suffers from epilepsy. Published articles have focused so far on evaluating changes in adult patients. The aim of the study was to evaluate anatomic structures’ volume of the brain in pediatric patients with epilepsy.

A group of 42 pediatric patients with clinical symptoms of epilepsy (study group) and 16 healthy patients (experimental group) aged 3 months-17 years were enrolled in the study. Brain MR imaging was performed in all children according to a dedicated protocol (epilepsy specific protocol). Individual anatomical structures of the central nervous system were analyzed on the basis of T1-weighted 3D isometric 1 mm sequence and volume changes of specific structures were compared between the epilepsy group and the control group.

In the study group, the ratio of brain tissue to CSF was 89,08% to 10,92%, while in the control group it was 90,99% to 9,01%. In the research group compared to the control group, the volumes of each brain structure were: cerebrum – 77,99%/79,68%, cerebellum – 9,65%/9,87%, brainstem – 1,41%/1,44%, lateral ventricle – 1,41%/0,49%, caudate – 0,54%/0,55%, putamen – 0,61%/0,68%, thalamus – 0,82%/0,91%, globus pallidus – 0,18%/0,19%, hippocampus – 0,45%/0,50%, amygdala – 0,11%/0,10%, accumbens – 0,04%/0,05%.

During the course of epilepsy in pediatric patients, there is a decrease in the volume of brain tissue, with particular emphasis on the cerebrum, cerebellum, brainstem, caudate, putamen, thalamus, globus pallidus, hippocampus and accumbens, moreover an increase in the volume of lateral ventricles. The study indicates cortical and subcortical atrophy in pediatric patients with epilepsy. The data obtained have important clinical and prognostic significance, however they need to be confirmed on a large study group with taking into account changes in the volume of anatomical structures of the brain in relation to age and disease duration.