# TUBEROUS SCLEROSIS (CLINICAL CASE)

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**Relevance**. Tuberous sclerosis is a rare genetic disorder that causes a tendency for benign tumors to develop throughout the body. The incidence rate is one case per 30 thousand population. Depending on the organ in which the pathological focus is localized, the patient may experience a varied clinical picture. Damage to the brain and spinal cord in tuberous sclerosis are the most common.(1,2)

**Purpose.** Determine the role of a multidisciplinary approach in the diagnosis and treatment of tuberous sclerosis

**Materials and research methods**. A pro- and retrospective analysis of the medical card of an stationary patient who was in the Department of Neurology of the NCMC was carried out. Patient: P.S. Age: 10 years old . Complants on admission: epileptic attacks in the form of serial "nods", fading, adversion of the eyes to the side, orofatial automatisms ( duration up to 20 seconds, post-attacking period-sleep up to 2 hours). The department conducted a complete in-depth examination: laboratory (general blood and urine test, biochemical blood test, general stool analysis, molecular genetic examination) and instrumental tests (brain MRI, ECG, EEG, internal ultrasound, ECHO-KG). The child was examined by narrow specialists: ophthalmologist, neurologist

**Results and discussion**

Anamnesis of the disease

The debut of attacks in 1 year. After 30 minutes after falling asleep, she opened her eyes, unconscious, the orofacial component (grimace) lasting up to 1 minute, the frequency is single per day. EEG-without epileptiform activity. Depakin is introduced into therapy. Rare attacks persisted once every 3-6 months, the AEDs dose was corrected. Since March 2016, Trileptal has been introduced into therapy, against this background, a remission of 9 months has been achieved.

OBJECTIVE EXAMINATION

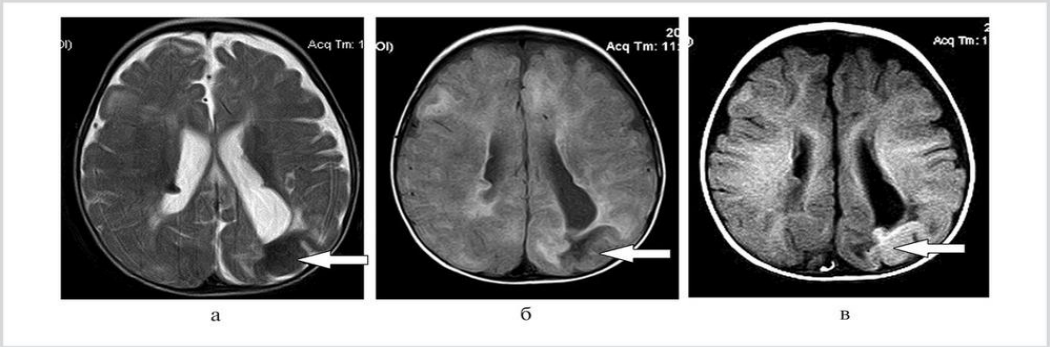
Somatic status. Medium weight. The skin is clean. There are multiple hypopigment spots on the torso and limbs. Areas with an uneven "shagren leather" texture. Angiofibromas on the face. Colon-nail fibroma on the middle toe of the left foot. On the left cheek there are several flat formations with somewhat thickened skin, pink colour. Visible mucous membranes are clean, moderate humidity. The yen is calm, there are no plaques. Lymph nodes are not enlarged. The tonsils are not enlarged. The breath is vesicular. The tones of the heart are clear, rhythmic. Functional noises: no. HR 87 per minute. The stomach is soft, painless. The liver is not enlarged. The chair is decorated, without pathological impurities. Body weight (kg): 41

EXAMINATION AT THE DEPARTMENT

- Brain MRI: MR picture of tuberous sclerosis, with multiple tubers and subependymary nodes, without signs of Monroe's opening occlusion.
- ECG: sinus rhythm. Sinus tachycardia.

- EEG: generalised epileptiform changes with anterior projection predominance are registered. The index of epileptiform activity varies up to 40%
- Abdominal ultrasound: diffuse changes in kidney parenchyma
- ECHO KG: rabdomyoma resorption in intraventricular septum
- Ophthalmologist: Simple hypermetropic astigmatism of both eyes. Retinal hamarthroma of both eyes
- Molecular genetic study : mutation in the TSC2 gene was detected
- Clinical and laboratory control: no change

MRI of the brain of a child with TC , axial sections: cortical tuber (arrow). a - T2-weighted image; b - FLAIR-image; c - T1-weighted image.




DIAGNOSIS: Main : Tuberous sclerosis (Subependimal giant cell astrocytoma of the right lateral ventricle of the brain. Accompanying: Localised (focal) (partial)symptomatic epilepsy and epileptic syndromes with complex partial seizures (Structural focal epilepsy, pharmacoresistant form. Multiple kidney angiolipomas. OU gamarthromas)

Therapy. Vigabatrin 2000mg 2 times a day (4000mg/day), depakin chronosphere 450mg morning, evening (900mg/day), ethosuximide 250mg morning, evening, levetiracetam 8 ml in the morning and evening, affinitor 10mg morning

**Conclusion**

Managing patients with symptomatic epilepsy in TSC requires a multidisciplinary approach. Early diagnosis and timely adequate therapy of epilepsy in TC improve the prognosis of the disease and the quality of life of patients in the future.

## References:

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2. Curatolo P , Maria BL. *(https://pubmed.ncbi.nlm.nih.gov/23622183/). Handb Clin Neurol*. 2013;111:323-331. Accessed 1/5/2023.