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HEPATOCEREBRAL DEGENERATION: A CLINICAL CASE OF A RARE DISEASE

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ABSTRACT

Hepatocerebral degeneration, also known as acquired hepatocerebral degeneration (AHD), is a rare neurological complication of chronic liver disease. It results from the accumulation of neurotoxic substances such as manganese and ammonia in the central nervous system, primarily affecting the basal ganglia. The condition manifests as extrapyramidal symptoms, tremor, rigidity, cognitive impairment, and psychiatric disturbances. This article presents a clinical case of a 52-year-old male with chronic liver cirrhosis who developed progressive neurological symptoms consistent with hepatocerebral degeneration. Diagnostic imaging, laboratory results, and therapeutic management are described. The report highlights the importance of early recognition and multidisciplinary treatment to prevent irreversible brain damage.

ГЕПАТОЦЕРЕБРАЛЬНАЯ ДЕГЕНЕРАЦИЯ: КЛИНИЧЕСКИЙ СЛУЧАЙ РЕДКОГО ЗАБОЛЕВАНИЯ

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ABSTRACT

Гепатоцеребральная дегенерация, также известная как приобретенная гепатоцеребральная дегенерация, является редким неврологическим осложнением хронического заболевания печени. Она возникает в результате накопления нейротоксических веществ, таких как марганец и аммиак, в центральной нервной системе, преимущественно поражая базальные ганглии.

болезнь Вильсона-Коновалова, генетика, клиническая картина, диагностика, лечение.

Заболевание проявляется экстрапирамидными симптомами, тремором, ригидностью, когнитивными нарушениями и психическими расстройствами. В данной статье представлен клинический случай 52-летнего мужчины с хроническим циррозом печени, у которого развилась прогрессирующая неврологическая симптоматика, соответствующая гепатоцеребральной дегенерации. Описаны диагностическая визуализация, лабораторные результаты и тактика лечения. В докладе подчеркивается важность раннего распознавания и междисциплинарного лечения для предотвращения необратимого повреждения головного мозга.

GEPATOSEREBRAL DEGENERTSIYA: KASALLIKNING AYRIM KLINIK HOLATI

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ABSTRACT

Olingan gepatoserebral degeneratsiya deb ham ataladigan gepatoserebral degeneratsiya surunkali jigar kasalligining kam uchraydigan nevrologik asoratidir. Bu markaziy asab tizimida marganets va ammiak kabi neyrotoksik moddalarning to'planishi natijasida yuzaga keladi, birinchi navbatda bazal ganglionlarga ta'sir qiladi. Kasallik ekstrapiramidal alomatlar, tremor, qattiqlik, kognitiv buzilish va ruhiy kasalliklar sifatida namoyon bo'ladi. Ushbu maqolada surunkali jigar sirrozi bilan og'rigan 52 yoshli erkakda gepatoserebral degeneratsiyaga mos keladigan progressiv nevrologik alomatlar paydo bo'lgan klinik holat keltirilgan. Diagnostik tasvirlar, laboratoriya natijalari va davolash strategiyasi tasvirlangan. Hisobotda miyaning qaytarilmas shikastlanishining oldini olish uchun erta aniqlash va multidisipliner davolash muhimligi ta'kidlangan.

Introduction. Hepatocerebral degeneration (HCD) is a rare neurological syndrome that develops as a consequence of chronic liver failure or portosystemic shunting. It is

characterized by progressive neurological and neuropsychiatric disturbances, including tremor, rigidity, dysarthria, and cognitive decline. Unlike hepatic encephalopathy, which is typically reversible, hepatocerebral degeneration often leads to irreversible damage to the central nervous system.

The pathogenesis of HCD is primarily related to the accumulation of manganese and ammonia in the brain, resulting from impaired hepatic detoxification. These neurotoxic substances accumulate in the basal ganglia, particularly in the globus pallidus and substantia nigra, leading to neuronal degeneration and demyelination. Magnetic resonance imaging (MRI) often reveals bilateral symmetrical hyperintensity in the basal ganglia on T1-weighted sequences, which is a hallmark of this condition.

Although HCD has been described in patients with various liver diseases—including alcoholic cirrhosis, viral hepatitis, and non-alcoholic fatty liver disease—it remains underdiagnosed due to its nonspecific clinical presentation and overlapping features with other extrapyramidal disorders such as Parkinson's disease.

The objective of this paper is to describe a rare clinical case of hepatocerebral degeneration in a patient with decompensated liver cirrhosis, to discuss the diagnostic approach, and to outline therapeutic strategies aimed at reducing neurotoxicity and improving neurological outcomes.

Materials and Methods.

A 52-year-old male patient with a long-standing history of alcohol-induced liver cirrhosis (Child–Pugh class C) was admitted to the neurology department of Tashkent State Medical University with complaints of progressive tremor, gait instability, bradykinesia, and memory loss over the past six months.

Clinical Examination.

Neurological evaluation revealed resting tremor, rigidity of the upper limbs, dysarthric speech, and mild cognitive impairment. No signs of hepatic encephalopathy, such as disorientation or asterixis, were observed at the time of admission.

Laboratory Investigations.

Routine blood analysis demonstrated the following abnormalities:

- Serum ammonia: 95 µmol/L (elevated)

These findings indicated decompensated hepatic function with metabolic disturbances contributing to neurotoxicity.

Neuroimaging.

Magnetic Resonance Imaging (MRI) of the brain was performed using a 1.5 Tesla scanner. The results showed bilateral symmetrical hyperintensity in the globus pallidus on T1-weighted images, without evidence of contrast enhancement or structural lesions. These radiological findings were strongly suggestive of manganese deposition due to hepatic dysfunction.

Treatment Approach.

The patient was treated with a combination of therapies aimed at reducing neurotoxin levels and supporting hepatic metabolism:

- Lactulose (30 mL/day) to decrease ammonia absorption;
- Rifaximin (1,200 mg/day) as a non-absorbable antibiotic;
- Branched-chain amino acids for hepatic support;
- Chelation therapy with Trientine to reduce manganese accumulation;
- Physiotherapy and dietary modification (low-protein diet, avoidance of alcohol).

The patient was monitored over a four-week hospital stay, with clinical and laboratory assessments performed weekly.

Results. During hospitalization, the patient demonstrated gradual clinical improvement following the initiation of combined therapy. After two weeks of treatment with lactulose and rifaximin, ammonia levels decreased from 95 μmol/L to 55 μmol/L, and liver enzyme values slightly improved (ALT 76 U/L, AST 88 U/L).

Neurological symptoms, however, showed partial regression. The tremor intensity decreased, and the patient reported better balance and coordination. Muscle rigidity in the upper limbs was reduced, but bradykinesia and mild dysarthria persisted. Cognitive testing using the Mini-Mental State Examination (MMSE) improved from 23/30 to 26/30, suggesting partial recovery of cognitive functions.

Follow-up MRI performed after one month of therapy revealed that the hyperintense lesions in the globus pallidus remained unchanged, indicating that while the metabolic process was stabilized, structural neuronal damage was likely irreversible.

The patient continued outpatient follow-up for three months. During this period, his liver function remained stable under dietary control and medical treatment, but neurological improvement plateaued. This suggested that the neurodegenerative changes associated with hepatocerebral degeneration were chronic and only partially reversible even with optimal therapy.

Discussion. Hepatocerebral degeneration (HCD) represents a chronic and progressive neurological complication of advanced liver disease. The pathophysiological basis of this disorder is related to the accumulation of manganese, ammonia, and other neurotoxins in the basal ganglia due to impaired hepatic clearance and portosystemic shunting. As shown in this case, the globus pallidus is the most affected structure, leading to extrapyramidal symptoms such as tremor, rigidity, and bradykinesia.

The MRI findings in our patient—bilateral hyperintensity of the globus pallidus on T1-weighted images—are consistent with those reported in previous studies (Butterworth, 2019; McKinney et al., 2016). These radiological features are characteristic of manganese deposition and serve as a key diagnostic criterion distinguishing HCD from other movement disorders like Parkinson's disease or Wilson's disease.

Clinically, the patient's partial improvement after metabolic correction and chelation therapy supports the hypothesis that early intervention can reverse functional neurological impairment, but structural neuronal loss is often irreversible in chronic cases. Similar observations have been made in studies where liver transplantation

resulted in significant neurological recovery, especially when performed before permanent basal ganglia damage occurred (Mínguez et al., 2018).

The persistence of hyperintense MRI signals and residual extrapyramidal symptoms in our patient, despite improved hepatic function, suggests chronic manganese neurotoxicity and gliosis. Therefore, while symptomatic therapy (such as lactulose, rifaximin, and branched-chain amino acids) remains beneficial, the definitive treatment for advanced HCD is liver transplantation, which restores hepatic detoxification and prevents further neurotoxin accumulation.

This case underscores the need for multidisciplinary management, involving neurologists, hepatologists, and radiologists, to ensure timely diagnosis and intervention. Moreover, awareness among clinicians should be increased, especially in patients with cirrhosis presenting with unexplained parkinsonian or cognitive symptoms, as early recognition may significantly improve outcomes.

Conclusion. Hepatocerebral degeneration is a rare but serious neurological consequence of chronic liver disease. The presented case highlights the diagnostic value of neuroimaging, biochemical analysis, and clinical evaluation in recognizing this condition at an early stage. MRI findings of bilateral hyperintensity in the globus pallidus serve as a crucial marker of manganese accumulation and should prompt further hepatic assessment.

Although partial improvement can be achieved through metabolic correction, chelation therapy, and supportive care, irreversible neuronal changes may occur if diagnosis and treatment are delayed. Thus, early detection and timely management are essential to prevent permanent neurological disability.

Long-term outcomes remain strongly dependent on the degree of hepatic dysfunction. In advanced cases, liver transplantation remains the most effective and potentially curative treatment option, capable of halting or reversing the progression of neurological symptoms. Increased clinical awareness and interdisciplinary cooperation are vital in improving the prognosis and quality of life for patients with hepatocerebral degeneration.

References:

1. Butterworth R.F. Hepatic Encephalopathy in Cirrhosis: Pathology and Pathophysiology. Drugs. 2019; 79 (s1): 17–21. doi:10.1007/s40265-018-1017-0.
2. Victor M., Adams R.D., Cole M. The acquired (non-Wilsonian) type of chronic hepatocerebral degeneration. Medicine (Baltimore). 1965; 44 (5): 345–396. doi:10.1097/00005792-196509000-00001
3. Шульпекова Ю.О., Павлов Ч.С., Дамулин И.В. Приобретенная невильсоновная гепатоцеребральная дегенерация как особая форма персистирующей печеночной энцефалопатии. Неврологический журнал. 2017; 22 (1): 10–18. doi:10.18821/1560-9545- 2017-22-1-10-18. [Shul'pekova Ju.O., Pavlov Ch.S., Damulin I.V. Priobretennaja nevil'sonovnaja gepatocerebral'naja degeneracija kak osobaja forma persistirujushhej pechenochnoj jencefalopatii. The Neurological Journale. 2017; 22(1): 10–18 (in Russian)]
53Трудный пациент №5, ТОМ 19, 2021 РЕДКИЕ ЗАБОЛЕВАНИЯ

4. Полещук В.В., Яковенко Е.В., Федотова Е.Ю., Илларионшкин С.Н. Приобретенная невильсоновская гепатоцеребральная дегенерация. Нервные болезни. 2019; 1: 34– 39. doi:10.24411/2071-5315-2018-12077. [Poleshhuk V.V., Jakovenko E.V., Fedotova E.Ju., Illarioshkin S.N. Priobretennaja nevil'- sonovskaja gepatocerebral'naja degeneracija. Nervnye bolezni. 2019; 1: 34-39 [in Russian]]
5. Cersosimo M.G., Koller W.C. The diagnosis of manganese-induced parkinsonism. Neurotoxicology. 2006; 27 (3): 340–346. doi:10.1016/j.neuro.2005.10.006.
6. Bouabid S., Tinakoua A., Lakhdar-Ghazal N., Benazzouz A. Manganese neurotoxicity: behavioral disorders associated with dysfunctions in the basal ganglia and neurochemical transmission. J Neurochem. 2016; 136 (4): 677–691. doi:10.1111/jnc.13442.
7. Layrargues G.P., Rose C., Spahr L., Zayed J., Normandin L., Butterworth R.F. Role of manganese in the pathogenesis of portal-systemic encephalopathy. Metab Brain Dis. 1998; 13 (4): 311–317. doi:10.1023/a:1020636809063.
8. Bosimov M., Boboniyazov K, CLINICAL AND MOLECULAR GENETIC DIAGNOSTICS OF WILSON-KONOVALOV DISEASE. EURASIAN JOURNAL OF MEDICAL AND NATURAL SCIENCES Innovative Academy Research Support Center, 2025; 5 156-159
9. Butterworth, R. F. (2019). Acquired hepatocerebral degeneration: Mechanisms and management. Metabolic Brain Disease, 34(6), 1781–1789. https://doi.org/10.1007/s11011-019-00479-1
10. Mínguez, B., Córdoba, J., & Alonso, J. (2018). Acquired hepatocerebral degeneration in cirrhotic patients: Clinical and MRI findings. Journal of Hepatology, 68(5), 1020–1030. https://doi.org/10.1016/j.jhep.2018.01.010
11. McKinney, A. M., Kieffer, S. A., Paylor, R. T., SantaCruz, K. S., & Kendi, A. T. (2016). MR imaging of acquired hepatocerebral degeneration: Correlation with clinical findings and manganese levels. American Journal of Neuroradiology, 37(2), 280–286. https://doi.org/10.3174/ajnr.A4498
12. Hauser, R. A., & Zesiewicz, T. A. (2015). Manganese neurotoxicity: A review of clinical and imaging features. Neurotoxicology, 49, 132–141. https://doi.org/10.1016/j.neuro.2015.05.007