neuropeptides, metabolites values in treatment group compared to control.
Conclusions: Brain neurotransmitters play a vital role in brain functioning and also have important function in HD status. It remains to be examined the clinical efficacy of such neurotransmitters and to investigate in-depth the neural networks suggested in the extrapyramidal system. Thus, it can be concluded that restoring the neurotransmitters balance in the brain may prevent or delay the symptoms of movement disorders.

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STUDY OF THE DIFFICULTIES OF LATE DIAGNOSTICS IN PATIENTS WITH HUNTINGTON'S DISEASE IN UZBEKISTAN

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Background: To identify the causes that lead to late diagnosis of Huntington's disease.
Methods: To this study was included 17 patients with a confirmed molecular genetic diagnosis, followed up with us from 2019 to the present.
Results: According to our observations, the disease often debuts with mental disorders (3 families), and therefore these patients are observed in psychiatric hospitals and examined by neurologists and geneticists out of time or do not receive consultations from these specialists at all. We have described a patient with Huntington's disease, in whom the disease debuted with cranial dystonia. In 2 families, the debut of the disease in the son is observed earlier (at the age of 35 years) and more severe than in the father (at the age of over 70 years) - the phenomenon of anticipation. An allele with incomplete penetrance (36–38 repeats) was identified in 2 families.
Conclusions: The main reasons leading to late diagnosis of Huntington's disease are identified: 1) The possibility of disease manifestation with cranial dystonia or other atypical neurological symptoms; 2) The presence in the population of alleles with incomplete penetrance, which complicates the early diagnosis of the disease due to the frequent absence of a family history, in some cases - the minimum severity of choreic hyperkinesis and intact intelligence; 3) The phenomenon of anticipation, leading to diagnostic errors, since the disease debuts in parents later than in children, and has an erased clinic; 4) Manifestation of the disease with mental disorders and frequent cases of suicide in the family before the start of a typical clinic, which leads to long-term observation by psychiatrists.

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"LEVINE-CRITCHLEY SYNDROME" - OBSOLETE

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Background: In the 1960s, families of patients with neurological features and red blood cell acanthocytosis in the absence of lipoprotein abnormalities were described independently by Irving Levine and Edmund Critchley (PMID 5677189; 5636069). The condition was later summarized under their names (OMIM #200150), alternatively as "neuroacanthocytosis", "chorea-acanthocytosis/choreoacanthocytosis", "familial tic disorder, parkinsonism, motor neuron disease, and acanthocytosis" and "amyotrophic chorea with acanthocytosis". In 2011, we showed that Critchley's original family from Kentucky was affected by mutations in the autosomal VPS13A gene (PMID 21987550).
Methods:DNA from the asymptomatic daughter of Levine's index case (one of a pair of brothers) was analysed.
Results: We confirmed the presence of a mutation in X-chromosomal gene XK, that was previously found in a distant, asymptomatic female relative who had contacted us for advice on her family's condition.
Conclusions: We conclude that the term "Levine-Critchley syndrome" applies to genetically diverse conditions and is no longer of medical value. It must be replaced by proper molecular diagnoses of "VPS13A disease" or "XK disease", respectively. It is not surprising that the similarity (and thus confusion) of these ultra-rare syndromes results from molecular interaction of the two respective proteins (PMID 32845802). Clinically, the distinct genetic conditions may be distinguished on the basis of Kell blood group typing (McLeod phenotype in XK disease), heart exam (XK: severe involvement), recessive (VPS13A) or X-linked (XK) inheritance, and less so on different ages of onset (VPS13A: young adult males and females, often siblings; XK: elderly males, often brothers or nephew-uncle pairs, but only exceptionally manifesting in females).

Part III M: Ataxias, hereditary spastic paraparesis
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LATE-ONSET CEREBELLAR ATAXIA: CASE REPORT OF A NEW CNV ON *TTBK2* GENE AS POSSIBLE CAUSE OF SCA-11

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Background: The spinocerebellar ataxias (SCAs) are a group of neurodegenerative diseases that affect not only the cerebellum but also other nervous system structures such as brainstem, spinal cord, or peripheral nerves. According to Harding's classification, SCAs are classified into three types. Type 3 of SCAs include pure autosomal dominant cerebellar ataxias, the most common group of inherited ataxias. However, in some cases, this type of pure ataxia could coexist with other clinical signs such as tremors, abnormal eye movements, or pyramidal signs. The main cause is a trinucleotide expansion which encodes polyglutamine proteins; however, others can be caused by missense mutations or small insertion/deletion variants. SCA-11 is included in type 3 of SCAs and accounts for less than 1% of autosomal dominant ataxia in Europe. The pathogenic variant involved, is the *TTBK2* gene.
Methods: We present a case of an adult patient with a SCA, followed by a genetic analysis.
Results: A 68-year-old woman presented with a 10-year history of imbalance and abnormal gait. The patient was previously healthy, and her medical and familiar history was irrelevant. On the physical exam, she showed signs of cerebellar ataxia and hyperreflexia in lower extremities. An approach to exclude acquired causes of cerebellar ataxia was performed, including complete serum blood count, metabolic panel, liver function test, thyroid function tests, vitamin B12 and B1, anti-GAD, antigliadin, anti-endomysial and anti-transglutaminase antibodies, autoimmune panel, all of them without any abnormality. The brain MRI showed brainstem and cerebellar atrophy. Exome sequencing, copy number variants, and mitochondrial genome tests were performed. A copy number variant 43008859_43075833 on the *TKKB2*gene was detected on chromosome 15 in the genomic location of GRCh37.
Conclusions: We report a case of a copy of number variant not previously reported as pathogenic on *TKKB2*gene that can be the cause of spinocerebellar ataxia in our patient.

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SPASTIC ATAXIC PHENOTYPE OF FATTY ACID 2-HYDROXYLASE ASSOCIATED NEURODEGENERATION: A CASE SERIES FROM INDIA

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Background: Fatty acyl 2-hydroxylase (FA2H) neurodegeneration (FAHN) is a type of neurodegeneration with brain iron accumulation (NBIA) that presents with pyramidal, extrapyramidal, cerebellar features and cognitive