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Neuro-gastrointestinal encephalopathy (MNGIE) as mitochondrial dysfunction: Difficult diagnosis and effective treatment

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MNGIE is a mitochondrial disease that is characterized by disorders of the nervous system, endocrine system and the gastrointestinal tract. We have gained experience of prenatal manifestation of the disease. The diagnosis of which was established at the age of 19. In other cases, the disease manifested at 33 and 27 years.

The purpose of the study is to study the frequence and the nature of the clinical manifestations of a rare hereditary disease - MNGIE.

Materials and methods: classic clinical-genetic and modern technologies are used. Patient S. referred with a diagnosis of colitis, malabsorption syndrome, gastroduodenitis, cachexy.

Results of the study: Complaints of progressive weight loss (30 kg for 7 years), epigastric pain, flatulence, altered defecation pattern (alternating constipation with diarrhea), severe general weakness, amenorrhea.

Ill since 7 years: epigastric pain, progressive weight loss. After the flu, the condition progressively worsened: pain in the epigastrium, general weakness, weight loss increased. Pedigree is burdened with neurological (hyperkinetic syndrome) and multifactorial pathology (hypertension, chronic gastritis). Height - 166 cm, weight - 35 kg, severe cachexia, mongoloid eye incision, divergent strabismus, protruding ears, thoracic spine scoliosis, chest deformity, hyperemia of nose skin, hands, feet, moderate myopia, astigmatism. In the neurological status - ataxia, tremor.

Biochemistry: ↓ glucose, ↓ total protein, ↓ calcium, ↓ folic acid.

Ultrasound: Moderate diffuse changes in the liver parenchyma, low location of the gallbladder, signs of pancreatopathy, crystaluria.

The course of individual rehabilitation drug therapy, which primarily included energy therapy, as well as metabolic cofactor therapy and diet therapy:

- Coenzyme Q
- cytochrome C
- L-carnitine
- creon
- omeprazole
- probiotics
- microhydrin
- vitamins B6, C, E, folic acid

Results: In the case of MNGIE, it is possible to avoid death by diagnosing and effective treatment.

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