Khomenko M.A., Strelkova M.I, Omelchenko O.V.

Rare clinicalcase: syndrome HHH  
Kharkiv National MedicalUniversity  
Department ofPediatrics No. 1 with Neonatology, Kharkiv, Ukraine  
Supervisor – prof. Senatorova A.  
   
  
Introduction. Syndrome HHH (hyperornithinemia, hyperammonemia, homocitrullinuria)is an orphan hereditary disease associated with defect of ornithine transporterand manifested by high level of ammonium and ornithine ions in blood along withincreased renal excretion of homocitrulline. The disease is caused by agenetically determined defect of ornithine transporter.Syndrome is characterized by variable age of onset: from the neonatal period to 18 years. The disease is paroxysmal.Early symptoms are often not specific, therefore they cannot be easily recognized. Initial clinical symptomsand regular attacks of illness are provoked by infection, anesthesia,artificial feeding, high-protein food.  
  
Purpose. Improving diagnostic of orphan diseases.  
  
Methods. Clinical observation of the 4 y.o. child S. with thesyndrome HHH. Polymorphism in a system of genes of a folate cycle.Hyperhomocysteinemia. Metabolic encephalopathy, the III level brain coma.  
  
Results. Justification of diagnosis: frequent vomiting,weakness, lethargy, delay in psycho-speech development, muscle twitching,somnolence episodes; anesthesia and faults in the diet in anamnesis; familyhistory (father has unwarranted aggression periods, rapid attention depletion,hyperactivity);vestibular-ataxic syndrome; attention deficit hyperactivity disorder;  
 liquor-venous discirculation;  
increaseof cytolysis in 7 times, 10-times ammonium increase in blood, 1,5-timesornithine increase, 1,2 times citrulline increase; progressive course of the disease; multi-organfailure development.  
  
Conclusions. Hereditary metabolic disorders must be excludedwhile identifying the neurological symptoms of the unknown etiology. Patientsshould be examined comprehensively involving geneticists. That helps todiagnose hereditary metabolic disorders on early stages, and therefore, preventcomplications.