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A case of mixed geno—Phenotype of generalized dystonia and strumpel disease*

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ABSTRACT

Background: Strumpel disease and dystonia are inherited disorders with the clinical picture of spastic paraparesis and hyperkinesis respectively. We present a case of a patient born from parents with these diseases who developed neurologic phenomena uncharacteristic for the classical clinical picture of his parents' disorders. Case report: Patient V., 12, born from his father with generalized dystonia and mother with Strumpel disease, has flaccid lower paraplegia along with dystonic hyperkinesis in neck and arms. Discussion: The flaccid lower paraplegia could be caused by the anterior horn lesion. This phenomenon is unclear because anterior horn lesions were not diagnosed in the proband's parents.

Keywords: Dystonia; Strumpel Disease; DYT 1

1. INTRODUCTION

Dystonia and Strumpel diseases, *i.e.* hereditory spastic paraparesis (HSP), are heterogenious conditions; mutations in more than 50 different genetic loci have been described to cause HSP and more than 20 different genetic forms of dystonia. Lesions of extrapyramidal system are responsible for clinical signs of dystonia with extrapyramidal rigidity in muscles. In case of Strumpel disease with lower spastic paraparesis, caused by lesions of corticospinal tract, spasticity in muscles grows by a pyramidal type. Flaccid paresis is not characteristic for either of these two disorders. Patient V., 12, born from his father with generalized non-DYT 1 dystonia and mother with Strumpel disease, has flaccid lower paraplegia along with hands and neck dystonia.

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2. CASE STORY

I first met patient S. with generalized non-DYT 1 dystonia in 1987 on summer practice at neurologic department, being a third year student of a medical school. He then was 21. He fell ill at the age of 9 and soon involuntary movements became generalized. None of his relatives was affected by this disease. Though classical research of Eldridge [1] established superior IQ of patients with recessive forms of dystonia, we observed very clever and vivid mind in our patient. Later he married a woman with indolently progressive Strumpel disease, and their son inherited both parents' diseases—from early childhood he developed flaccid lower paraplegia with complete absence of strength in legs and dystonic hyperkinesis in neck and arms, while having IQ as high as his father's. The son underwent embryonal transplantation surgery into his brain with no significant effect. His father—patient S.—when hospitalized was given L-Dopa with no effect. I started tryhehyphenidyl therapy for patient S, which resulted in considerable improvement in his condition. Test for DYT 1 proved negative. Along with test results of my other primary dystonia patients from the monitored group his serotonin exchange tended to be on the increase. His videoexamination was conducted before tryhehyphenidyl therapy (**Figures 1-3**). Unfortunately, the mother of the proband refused to videotype her son after brutal murder of her husband—patient S. on May 9th 2005—Russia's national Victory Day holiday; patient S. ventured to leave the house in his wheelchair for the first time after the winter and never got back. Later he was found dead with tens of stab wounds and his neck cut.

3. DISCUSSION

More than 20 genetic loci of dystonia have been discovered so far [2], but the clinical picture of the disorder of our proband's father, *i.e.* non DYT-1 early onset generalized autosomal dominant dystonia, differs from all

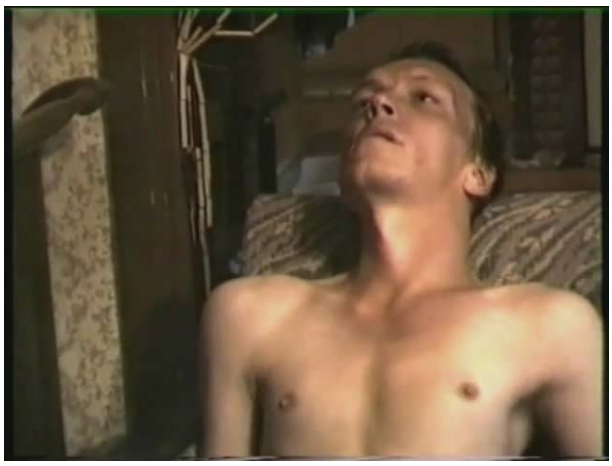

Figure 1. Patient S., father of proband, patient V., speaking.

Figure 2. Patient S., father of proband, patient V., sitting.

Figure 3. Patient S., father of proband, patient V., walking.

known phenotypes. The closest one is DYT-6 phenotype with gene mutated on chromosome 8, encoding apoptosis-associated protein, but in this type of dystonia legs are rarely affected. On the other side, Fasano *et al.*, who studied early-onset non-DYT-1 dystonia, revealed lower limbs' lesion in 37% of patients from this group [3]. The above mentioned facts together might be indicative of a possibility that the genetic defect, responsible for dystonia in our family and probably in some of Fasano's patients, differs from the 20 ones described so far.

Moderate flaccid paraparesis caused by partial loss of anterior horn is rarely observed in some forms of HPS [4]. However, total lower flaccid paraplegia can be explained by severe lesions of anterior horns. No such cases have ever been published before either for Strumpel disease or for dystonia. We can suggest with great caution that in this type of dystonia after loss of pyramidal control cell-defects in anterior horns are displayed in the form of flaccid paraplegia.

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