

Natureza do trabalho: Relato de caso

TÍTULO

WOLFRAM SYNDROME: A CASE REPORT

NATÁLIA DE CARVALHO AFONSO, RONAN MOUGENOT BELMONTE, MARCIA RIBEIRO RODRIGUES ANDRADE, ELIANE ARAÚJO, DAYANE DA SILVA LIMA, SÔNIA REGINA JURADO

UNIVERSIDADE FEDERAL DE MATO GROSSO DO SUL, UFMS, TRÊS LAGOAS, MS, BRASIL

RESUMO

Introduction: Wolfram syndrome (WS) is defined as a neurodegenerative condition involving the central and peripheral nervous system and neuroendocrine tissues. It is a rare, progressive, autosomal recessive, which was first described in 1938 by Wolfram and Wagner, whose incidence is estimated at 1:770.000 live births. **Case report:** Male patient, 12 years old, 32 kg and 1.30 m tall, a resident of the state of São Paulo, Brazil, with diabetes mellitus type 1 since the first year of life and started four years with progressive visual loss and auditory acuity. On physical examination the patient was hemodynamically stable, with low weight and short stature, delayed puberty, urinary incontinence and neurological abnormalities (hyperactivity, aggression and agitation). A family history of consanguinity, the parents being first cousins. Diagnostic laboratory tests were glycosylated hemoglobin. The clinical and laboratory tests confirmed 99% SW. The treatment is a medical palliative of physiological and psychological changes (Lantus, Humalog and Respiridona). Currently the teenager remains stable and performing activities normally. **DISCUSSION:** The present study review of the literature has shown that the clinical course of SW is variable, with the possibility of several syndromes, at the same time, they share some common clinical features classically described (diabetes mellitus associated with optic atrophy). The psychological support to patients and their families is essential due to the high morbidity and mortality of this disease. This support is also important due to changes in behavior and psychiatric disorders that the patient may develop over time. **Conclusion:** It is possible that the lack of health professionals about the association between diabetes mellitus and monogenic syndromes could be a major contributor to the underdiagnosis of diseases such as SW. In the patient reported, the relevant clinical manifestations are consistent with the average age of onset of complications.