OCCURRENCE OF SANFILIPPO SYNDROME IN TWO SISTERS

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ABSTRACT

This case report highlights a genetic disease, the Sanfilippo Syndrome type B, a genetic disorder characterized by an inborn error of metabolism. The incidence is around 1: 10,000 to 1: 25,000 newborns. The syndrome affects the central nervous system, having been reported its occurrence in two sisters.

KEYWORDS: Sanfilippo syndrome, autosomal recessive inheritance.

1. INTRODUCTION

The Sanfilippo syndrome type B (MPSIIIB) is a genetic disorder (17q25) characterized by an inborn error of metabolism caused due to deficiency of the enzyme a N-acetyl-glucosaminidase (NAGLU), affecting the processing of the glycosaminoglycan (GAG) heparan sulfate (HS)[1,2]. It was first described in 1963 by American pediatrician Dr. Sylvester Sanfilippo[2]. The incidence is around 1:10,000 to 1:25,000 newborns[3,4]. It originates from an autosomal recessive inheritance and has its clinical expression around 4 years of age, with progressive deterioration of cognitive skills, communication and behavioral[2,5]. It is a disease of permanent and progressive symptoms whose manifestations are multisystemic, affecting the central nervous system, skeletal, digestive, and often, the eye and the epithelial[2,5,6,7].

We carried out the report of this rare clinical case the of two sisters through a descriptive study. Data were collected from the medical records of patients, history, interviews with family members and caregivers. Both find themselves in therapeutic residence in an institution where some of the authors work.

For scientific technical background were searched the medical literature and health sites PubMed and Bireme between 1963 and 2014.

2. CASE REPORT

CASE 1 - Female, 36 years old, caucasian.

Assisted in Specialized Center institution Specialized Center Nossa Senhora D'Assunção since age 11. Dependent in ADL, uses wheelchair.

Absence of speech. Abnormal facial features. Dysostosis multiple. Restlessness, stereotyped hand movements, constant insomnia, oblivious to contact. Frowns, puts the tongue out of the mouth and makes faces all the time. Difficulty swallowing - uses thickener.

Medication used daily: Carbamazepine (400 mg), Pericyazine (10 mg), Clonazepam (1mg), Imipramine (25mg), Promethazine (50mg), Combiron (standard dose).

Diagnosis: Sanfilippo type B, Beta-thalassemia, Severe Mental Retardation, Epilepsy.

CASE 2 - Female, 27 years old, brunette.

Bone marrow transplantation was performed with 06 years old. Assisted in the institution Specialized Center Nossa Senhora D'Assunção since age 09.

Dependent in ADL, uses a wheelchair. Absence of speech. Dysostosis multiple. Restlessness, stereotyped hand movements. Sometimes addresses the look and features unmotivated laughter, screams and grimaces do all the time. Contact depleted. Difficulty swallowing - uses thickener.

Medication used daily: Neozine (25 mg), Haloperidol (2 mg), Clonazepam (2 mg), Tegretol (200 mg).

Diagnosis: Sanfilippo type B and Severe Mental Retardation.

In both cases, biochemical tests for the diagnosis were used to investigate the existence of enzyme deficiency. Radiological exams were also used for documentation of the case and composition of dignóstico (data not shown).

3. DISCUSSION

For the cases in question, both patients had development within the normal range with respect to sitting, crawling and walking. The speech has not been developed. They have medium height, although long bones and dorsal kyphosis. The losses in sphincters controls and gait started at around 05 years of age. The loss of ambulation happened gradually and before use of the wheelchair the march was true of the hip and knees bent. In both cases the use of the wheelchair occurred after 25 years.

Currently both have severe cognitive impairment affecting mainly the contact with the environment, with a deep alienation framework[6,7,8]. The syndrome causes significant neurological symptoms, including severe intellectual disabilities; the Intelligence Quotient may be less than 50. Symptoms appear more severe in people with Sanfilippo syndrome type A and yet there is no cure for individuals affected by any type of MPS III[6,7,8].

The Patient of Case 1 evolves with late-onset seizures culminating in increased use of anticonvulsants. Although studies and efforts in therapy, using transplants fibroblasts, bone marrow transplantation and enzyme replacement therapy, neither method is effective in remission of symptoms in order to prevent disease progression.

In the case of Patient Case 2, it is believed that the bone marrow transplant prevented the development of beta-thalassemia.

Both are under the care of a multidisciplinary team of professionals psychiatry, neurology, internal medicine, physical therapy, nursing, speech therapy, psychology, pharmacy and pedagogy. The performance of this team in the direct care and guidance of caregivers and family enables more effective control of the clinical variables culminating in improving the quality of life of patients as well as longevity of the cases, which according to medical literature, generally progresses with deaths in adolescence.

4. CONCLUSION

Based on the documentation of cases and literature data the authors concluded the diagnosis of Sanfilippo syndrome type B (MPSIIIB). In Case 2, it is believed that the bone marrow transplant prevented the development of beta-thalassemia. Scientists who study MPS III continue to search for better and more effective ways to treat them.

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