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CASE REPORT

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Stuve-wiedemann syndrome in a child: a case report

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Abstract

Introduction: Stüve-Wiedemann Syndrome was first described in 1971. However, this disease was considered a single entity only a few years later. The syndrome is now known to be an autosomal recessive disorder in which the patient has dysautonomia, bone dysplasia and respiratory distress and eating disorder. **Objective:** To report the case of a girl with Stuve-Wiedemann Syndrome. **Case Description:** A 7-year-old female SSG patient presented, at birth, with early neonatal sepsis, septic shock, pneumonia, respiratory acidosis, jaundice without the need for phototherapy, food intolerance, anemia, seizures, interstitial pulmonary edema, myositis and osteomyelitis of the right humerus. After 11 months of follow-up, she was referred to a maternal-fetal clinic for genetic testing to investigate the symptoms. The patient was diagnosed with Stüve-Wiedemann Syndrome. **Final Considerations:** This disease is considered a primary congenital bone dysplasia, characterized by skeletal and joint changes, bowing of the long bones, episodic hyperthermia, periodic respiratory infections, eating disorders and high mortality. These were facts found in the case of the child presented here. On the other hand, this disease can manifest other symptoms such as fissured tongue and episodes of hyperhidrosis, but it was not found in our patient's case. Rare diseases of genetic origin have a high negative impact on patients' quality of life. However, with the practice of physical activity, patients can have a healthy diet. Swimming practice by SSG presents itself as an extremely important activity for the physical and psychological development of this child, contributing to the improvement of her quality of life.

Keywords: Osteochondrodysplasias. Child syndrome. Case Report.

Introduction

Stuve-Wiedemann Syndrome was first described in 1971 in the work of Stuve A and Wiedemann HR, who observed two girls presenting congenital curvature of the femurs and tibias with abnormal foot positions. They also presented slight bowing of the long bones of the arms with hand anomalies and craniofacial and axial skeletal abnormalities. Both developed respiratory distress and eventually died a few days later in the neonatal period [1]. It demonstrates a condition that is influenced by genetic factors in the "long bone bowing" complex1. However, this disease was considered to be a single entity only a few years later [2].

The Syndrome is currently known as an autosomal recessive disorder, in which the patient has dysautonomia, bone dysplasia, respiratory distress and eating disorders [2]. Its prevalence is not yet known, but it is considered to be rare [3,4]. SWS treatment focuses on symptom management and supportive care because there is presently no cure for the condition [5]. The major types of treatment for people with SWS include orthopedic therapies to control bone abnormalities and enhance mobility, respiratory assistance for those who have breathing problems, and nutritional support to deal with feeding and growth challenges [5-7].

To further enhance quality of life, professional interventions like pain management, physical therapy, and occupational therapy may be suggested. Care for people with SWS also includes careful observation and control of potential side effects including scoliosis,

pneumonia, and cardiovascular problems [5,6,8,9]. It is also advised that affected people and their families get genetic counseling to better understand the hereditary basis of the condition and to go over family planning alternatives [5,9].

The cases of the disease are still scarce in the scientific literature, reporting the case of a new patient with the syndrome is extremely significant [10]. Therefore, the purpose of this work is to bring forward the case of a girl with Stuve-Wiedemann Syndrome.

Methods

Study Design

The present study was elaborated according to the rules of the CARE case report (https://www.care-statement.org/).

Ethical Approval

The study was submitted to the ethical committee with approval under CAAE number 52278821.5.0000.8083 and then the person in charge of the patient signed the free and informed consent form.

Case Report

A 7 year old female patient, SSG, was diagnosed with rare bone dysplasia called Stuve-Wiedemann Syndrome. She was born after 35 weeks, with 2340 grams and Apgar scores at 1 and 5 minutes were 8 and 9, respectively. The infant presented early neonatal sepsis with septic shock due to acute respiratory distress syndrome (ARDS) and pneumonia. During her hospitalization, she used CPAP for three days, moving to orotracheal intubation. She remained intubated for ten days, and presented extubation failure on the first attempt. She had respiratory acidosis, jaundice without the need for phototherapy, small atrial septal defect, food intolerance, seizures, anemia, interstitial pulmonary edema, and myositis with osteomyelitis. While in the hospital, a Guthrie test was performed along with eye and ear tests, which were within the parameters of normality. She also had taken BCG and hepatitis B vaccines.

Upon being discharged from the hospital, the patient was pale, hydrated, acyanotic, anicteric, and showed reddish and moist mucosas. There were no head trait abnormalities. The lungs had vesicular murmur without adventitious sounds. Precordium with normophonetic sounds in two beats without murmurs. Flaccid abdomen, liver 1 cm from the right coastal margin and hydro-air sounds present. Palpable pulses in the 4 limbs, extremities warm with two seconds of capillary refill. Upper limbs with vicious presentation of closed hands, fingers curled up. Lower limbs without anatomical deformities. Typical female genitalia. After in-hospital treatment, she was discharged with guidance for reassessment within 48 hours of return with a pediatrician, and outpatient follow-up with an orthopedist.

After 11 months of follow-up, she was referred to a maternal-fetal clinic to perform a genetic test to investigate the symptoms presented. The patient was diagnosed with Stüve-Wiedemann Syndrome (SWS).

During outpatient follow-up, in 2018, routine exams were performed, namely: urine I; urine culture and antibiogram; complete blood count; dosage of 25-hydroxyvitamin D, ferritin, glucose, TSH, T4 free, full cholesterol exam, HDL cholesterol, LDL cholesterol according to the Friedewald formula, triglycerides; hip radiography; echodopplercardiogram (ECO); and electrocardiogram.

The blood count showed hypochromic microcytic anemia with anisocytosis, absolute neutrophilia, absolute eosinophilia, absolute basophilia, relative and absolute lymphocytosis, absolute monocytosis, without platelet changes. On ECHO, there was moderate dilatation of the ascending aorta from the aortic valve to the emergence of the brachiocephalic trunk, and in the exams performed before, there was no significant change. The ECG showed no changes in heart rate or rhythm. On the hip radiography, dislocation of the hips, deformity of the proximal femurs with separation of the epiphyses and calcification in soft tissues were reported. The other exams were within the normal range.

The patient is currently monitored by a physical therapist, who helps her to perform physical activities, such as swimming. This has proven to be effective for the patient's neuropsychomotor development.

Discussion

Pathophysiologically, it is caused by a mutation of the leukemia inhibitory factor receptor (LIFR; 151433) gene on chromosome 5p13 [11,12]. Stüve-Wiedemann syndrome, also called type II Schwartz-Jampel syndrome, is considered a primary congenital bone dysplasia. It is characterized by skeletal changes, severe osteoporosis, joint contractures, camptodactyly, lower limb bowing, episodic hyperthermia, respiratory infections episodes, eating disorders and high mortality [4,13]. All listed in the case of the referred child.

Furthermore, there are reports that this syndrome could be associated with fissured tongue and episodes of hyperhidrosis, the latest being due to hair follicles anomalies, or an acquired follicular occlusion [13]. It is emphasized that facial dysmorphia is evidenced by blepharophimosis, flattened face and micrognathia

[14]. However, these alterations are not in agreement with the child here reported.

Some symptoms of SWS are very similar to those observed in Crisponi syndrome (CS), which is distinguished by the absence of osteochondrodysplasias [15]. According to Dahoneau N et al., Stuve-Wiedemann Syndrome is not always lethal in the neonatal period [15]. Diseases usually associated with this syndrome are myopia, strabismus, myelopathies, malignant hyperthermia, Von Willebrand's disease and mental development delay [16]. There is a higher risk for SSG to develop these diseases compared to the general population.

The diagnosis is typically collected through clinical and radiological examination after birth, however, as it is a rare disease, many doctors do not associate the infant's body alterations with SWS, therefore delaying the correct diagnosis [9,16]. Although genetic testing is not routinely performed, it can confirm the diagnosis of SWS [9,16,17]. Other methods can be used to diagnose the disease, such as electromyography and muscle biopsy, which does not have much evidence in the literature [17].

Though there are not many reports on the relationship between SWS and the quality of life of patients, according to Schieppati et al (2008) [18] and Forestier-Zhang et al (2016) [19], rare diseases of genetic origin have a high negative impact on patients' quality of life. Swimming practice presents itself as an extremely important activity for the neuropsychomotor development of SSG, contributing to the improvement of her quality of life.

Final Considerations

SWS is a rare genetic disorder caused by a mutation of the leukemia inhibitory factor receptor gene and is a primary congenital bone dysplasia characterized by skeletal change, severe osteoporosis, joint contractures, hyperthermia, respiratory infections and eating disorders. The diagnosis is typically made through clinical and radiological examination after birth. This disease also decreases the quality of life and swimming practice has a positive impact in quality of life and may contribute to improve their neuropsychomotor development. Despite being a case report with access only to the patient's initial exams from birth to the diagnosis of the syndrome, the patient's physical and motor follow-up could be observed. This case report will help many children diagnosed with this syndrome to improve their quality of life through swimming or even other physical activity.

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Ethical Approval

The study was submitted to the ethical committee with approval under CAAE number 52278821.5.0000.8083 and then the person in charge of the patient signed the free and informed consent form.

Informed consent

Not applicable.

Data sharing statement

No additional data are available.

Conflict of interest

The authors declare no conflict of interest.

Similarity check

It was applied by Ithenticate®.

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