Long-term follow-up of a 6-year-old girl with spinal muscular atrophy type 1. A case report

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ABSTRACT

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease characterized by the degeneration of alpha motor neurons in the spinal cord, leading to progressive paralysis and weakness of the proximal muscles. The disease affects between 1 in 6,000 and about 1 in 10,000 patients, and it is the most common hereditary cause of death in children. It is a severe, incurable, and progressive disease that leads to respiratory failure and immobilization for many patients. The authors present a long-term course of multi-specialist care in a girl with SMA type 1.

**Keywords:** Spinal muscular atrophy, SMA, type 1, neuromuscular disease

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INTRODUCTION

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease characterized by the degeneration of alpha motor neurons in the spinal cord, leading to progressive paralysis and weakness of the proximal muscles [1]. The cause of SMA is considered to be mutations in the SMN1 gene, which is 95% of cases consists of a homozygous deletion within the fifth chromosome (5q12.2-5q13.3), sometimes also has the character of a point mutation. In addition, in the case of 5% of patients, this condition may occur due to mutations in different genes called non-5q-SMA [2]. The clinical classification of spinal muscular atrophy is based on the age of onset and the maximum function achieved by the patient. It is essential to wait a certain period before assigning a patient to 1 out of 5 classes of disorder because numerous studies indicate that the maximum function achieved is more closely related to life expectancy than the age at the time of illness [3]. The disease affects between 1 in 6,000 and about 1 in 10,000 patients and is the most common hereditary cause of death in children [4]. SMA type 1 begins in infancy and contributes to the non-achievement of milestones or death and the absolute need for mechanical ventilation up to 2 years [5]. It is a severe, incurable, and progressive disease that leads to respiratory failure and immobilization for many patients. In addition, spinal muscular atrophy, despite the improvement in standards of care, is mentioned as one of the most common causes of death for children suffering from genetically determined diseases. What is more, SMA is also the most common neuromuscular disease diagnosed in infancy [6].

CASE REPORT

A girl, a child of the carrier’s father, and the carrier’s mother (homodeletion of exons 7 and 8 of the SMN1 gene and the presence of one copy of the SMN2 gene) were detected, carrying the mutation in both parents. The girl was born from a second, single pregnancy, born by cesarean section at 39 weeks, with a birth weight of 2840 g. The patient scored 2 points according to the Apgar scale. In addition, generalized hypotonia, popliteal contractures, and poor spontaneous respiratory activity were found at birth—clinically rated SMA 0/1. The clinical diagnosis of SMA was confirmed by a genetic study that showed genome imbalances manifested by a complete lack of signal for probes representing exons 7 and 8 of the SMN gene. However, one copy of exon seven and exon 8 of the SMN2 gene was found. In addition, the result of the molecular examination indicated the presence of extensive pathogenic deletion within the SMN1 gene, identified by the presence of exon seven deletions occurring in the homozygous system.

The study results confirmed the girl's clinical diagnosis of spinal muscular atrophy. From the beginning, the girl required assisted breathing. In addition, from birth, the patient was covered by HELP care at home (TRILOGY 100 respirator, oxygen concentrator, mammal, pulse oximeter, rehabilitation). The girl from birth was repeatedly hospitalized to treat chronic respiratory failure, frequent infections, and inflammatory changes in the lungs. In 2017, a 26-day-old girl was hospitalized for treatment for chronic respiratory failure. At admission, the general condition is severe, auscultatively over the pulmonary fields, alveolar murmur symmetrical, numerous bilateral furrows, disappearing after suction of mucous secretions from the respiratory tract. Due to the disease’s progressive nature, on 27 January 2017, a tracheostomy procedure was performed. The child's condition initially remained very severe. Attention was drawn to the increasing, massive peripheral edema response to intensive anti-edematous treatment. The child was consulted multidisciplinary. Due to the severe condition of the child and the lack of possibility of effective therapy, a conversation was conducted with the parents about not taking resuscitation activities in the event of cardiac arrest. During the 7-month stay in the ICU, the child twice experienced two episodes of deterioration of general condition with an increase in infectious parameters and inflammatory changes in the lungs. Antibiotic therapy was used following the obtained antibiogram.

On 29 June 2017, a gastrostomy procedure was performed. The postoperative course was good. Since birth, the girl has remained under the care of a neurologist. Neurologically, a child without muscular activity spontaneously - only eye and eyelid movements. In 2019, in a neurological examination, features of flaccid quadriplegia: lack of active movements, significant limitations in passive mobility in the limbs, areflexia, and muscular atrophy. The child was consulted cardiological and initially leaked through PFO and interventricular. Due to the elevated RR values, Captopril was given with good effect. In 2019 - features of the underlying disease, respirator, PEG. From birth, the child is chronically rehabilitated. In 2020, a physiotherapeutic assessment was carried out, which showed 0 points on the CHOP INTEND scale.

A girl is aged two and six months was admitted for intrathecal treatment with Nusinersen. Ten more doses were administered in the following months, with the last one being on 13 December 2021, when the girl was almost five years old. The patient also regularly attends the hospital to receive Spinraza, which is used to treat SMA. The girl remains under the multi-specialist care of the Following Clinics: Neurological, Genetic, Nutritional Treatment, Laryngology, Pediatric, Ophthalmology, Cardiology, Orthopedic, and Rehabilitation.

DISCUSSION

Type 1 SMA is the most severe form of spinal muscular atrophy. Type 1 SMA patients develop hypoventilation, recurrent lower respiratory tract infections, pulmonary aspiration, dysphagia, and
developmental impairment, usually also suffer from respiratory failure and die before the age of 2 [7]. In patients with SMA, due to the very complicated nature of the ailments, an interdisciplinary procedure involving numerous specialists from various fields is necessary [8]. Nusinersen is the first drug therapy approved to treat spinal muscular atrophy (SMA). Research shows improvement in motor functions in all types of SMA. Nusinersen is safe and effective. It has been proven that treatment with Nusinersen improves the quality of life in patients with SMA type 1 [9]. Apart from pharmacological treatment, the rehabilitation of patients is of great importance. Therapy plays an essential role in strengthening muscles, increasing range of motion, and reducing spasticity. Kinesitherapy, physical procedures are also used, including massages, treatments using heat, balneotherapy, and electrotherapy. In addition, some patients require specialist speech, swallowing, and chewing therapy. Treatment of respiratory complications and orthopedic equipment in orthoses or wheelchairs may also be unavoidable [10]. The course of multidisciplinary care improved the quality of life of the presented patient and undoubtedly contributed to the minimization of type 1 SMA symptoms.

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Conflicts of interest
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