Respiratory Rehabilitation Management in Adult Neglected Spinal Muscular Atrophy Type II Patient: A Case Report

Laras Hapsari,* Siti Chandra Widjanantie**

**Department of Rehabilitation Medicine, Faculty of Medicine, Universitas of Indonesia, Cipto Mangunkusumo Hospital, Jakarta, Indonesia*
***Department of Rehabilitation Medicine, Faculty of Medicine, Universitas of Indonesia, Persahabatan Hospital, Jakarta, Indonesia*

Abstract

Introduction: *Physical Medicine and Rehabilitation Specialists (Physiatrists) usually take care of patients with Spinal Muscular Atrophy Type II in an early childhood. The progression of respiratory dysfunction should be considered as the most disabling condition in patient with Spinal Muscular Atrophy Type II and need further respiratory rehabilitation approaches.*
This case describes an adult Spinal Muscular Atrophy Type II that is considered a late or neglected case as a lost case in the management of respiratory rehabilitation but his remain functional reserve must be optimized to reach a manageable respiratory outcome in daily living.

Keywords*: Spinal Muscular Atrophy Type II, respiratory rehabilitation*

Korespondensi: **Hapsari Laras**
E-mail: *larashapsari28@gmail.com*

Manajemen Rehabilitasi Respirasi pada Pasien Dewasa dengan *Neglected Spinal Muscular Atrophy* Tipe II: Laporan Kasus

Laras Hapsari,[*] Siti Chandra Widjanantie[**]

**Department of Rehabilitation Medicine, Faculty of Medicine, Universitas of Indonesia, Cipto Mangunkusumo Hospital, Jakarta, Indonesia*
***Department of Rehabilitation Medicine, Faculty of Medicine, Universitas of Indonesia, Persahabatan Hospital, Jakarta, Indonesia*

Abstrak

Pendahuluan: *Dokter Spesialis Kedokteran Fisik dan Rehabilitasi (Fisiatris) biasanya menangani pasien Spinal Muscular Atrophy Tipe II pada usia muda. Progresivitas disfungsi pernapasan pada penderita Spinal Muscular Atrophy Tipe II harus dipertimbangkan sebagai kondisi yang sangat berdampak disabilitas sehingga memerlukan program rehabilitasi respirasi lebih lanjut.*
Kasus ini menggambarkan Spinal Muscular Atrophy Tipe II pada usia dewasa yang telah neglected dalam penanganan rehabilitasi respirasi namun kapasitas fungsional yang tersisa tetap harus dioptimalkan untuk mencapai luaran respirasi fungsional dalam aktivitas sehari-hari

Kata kunci*: Spinal Muscular Atrophy Tipe II, rehabilitasi respirasi*

Introduction

Neuromuscular Disorder (NMD) is a disorder caused by an abnormality of any component of the lower motor neuron, which includes anterior horn cell, peripheral nerve, neuromuscular junction, or muscle. This disease affects children and adults with variable onset over the life span and often progressive with variable severity and rates of progression. Rehabilitation in NMD needs accurate diagnosis, natural history and impairment profile, prognostic information and anticipatory guidance for the near future for rehabilitation team, family, and patient.[1]

Spinal muscular atrophy (SMA) is a term used to describe a varied group of inherited disorders characterized by weakness and muscle wasting, secondary to degeneration of both anterior horn cell of the spinal cord and lower brainstem motor nuclei without pyramidal tract involvement. SMA causative gene defect was survival motor neuron (SMN) 1 gene, along with a disease-modifying gene (SMN 2). SMN protein is important to maintain the integrity of motor neurons, dendrites, and axons. Deficiency in SMN protein leads to inefficient assembly of the machinery needed for normal cell functioning and has damaging effects on motor neuron development and survival. Inheritance pattern was autosomal recessive with a 25% risk of disease with both carrier parents. International Consortium on SMA classification divided into four types of SMA.[1]

Case Discussion

A male patient, 32 years old, presented with muscle weakness that began since he was seven months old. The patient could not sit independently. From the age 10 months old until 7 years old, he underwent physiotherapy 3-4 times/week with stimulation to sit and to stand in a standing frame (prescribed by orthopedic) but no significant changes. He got dislocation of the right hip, but his parents did not want him to be operated. He began using a wheelchair for ambulation.

He continued education from kindergarten until high school in a regular school. His IQ test result was 124 (superior intelligence). At that high school, his scoliosis worsened. The orthopedic planned to have an operation, but his family refused.

At 19 years old, he can only move his right thumb and index finger. All of his Activity Daily Living (ADL) is fully dependent. He moves with a motorized wheelchair that is controlled by his thumb when going outdoor, and a regular wheelchair for in-house ambulatory. He did not go to college and choose to

attend courses.

From 22 to 30 years old, he frequently got hospitalized due to pneumonia and GERD. He had difficulty to swallow when exhausted, and sometimes experienced dypsnea during sleeping. Currently, patient had already had Non-Invasive Ventilation (NIV) to support his breathing. He used the NIV when he had shortness of breath (in this situation the O_2 saturation mostly was around 95%) usually in the morning and evening before night sleep, for about 30 minutes.

From the physical examination, the vital sign was normal, no tachypnea and O_2 saturation was 97% without oxygen support. The patient had flexion contracture and weakness in the upper and lower extremity (MMT 0 except right thumb 3), LMN neurologic sign (hypotonia and areflexia), pectus excavatum, low chest expansion, retraction in supraclavicular and sub-costae, gasping when taking inspiration, inadequate lip seal with drooling, hypotonic buccal muscle, relative macroglossia and scoliosis. From the capnograph, hypercapnia was presented with value of end tidal CO2 was 75. Peak Cough Flow in this patient is 210 Liter/minute. From thorax radiologic examination, patient had thoracolumbar scoliosis, C curved with Cobbs Angle 50 degrees. Patient undergoing Flexible Endoscopic Evaluation of Swallowing (FEES) examination reveal oropharyngeal dysphagia.

For respiratory muscle weakness problem, rehabilitation goals are to maintain adequate ventilation, optimize respiratory muscle strength and cough ability, minimize respiratory complications (hypercapnia, mucus retention, pneumonia, or respiratory failure). The programs are to educate the patient and caregiver how to prevent infection (hand hygiene, use mask, avoid contact with unhealthy people), regular use of NIV, airway clearance through cough assistance, oxygen saturation diary, respiratory vaccination (influenza, Hib, PCV), monthly lung function monitoring (EtCO2, PCF, PFR, MIP, MEP), and optimize energy conservation.

In a large series from Germany, of 104 cases classified as SMA II (sits alone, never walks), 98% survived to the age of 10 and 77% to the age of 20. Retrospective analysis of SMA type II patient by Farrar et al. also reveals that survival probabilities at ages 1, 2, 4, 10, 20, and 40 years were 100%, 100%, 97%, 93%, 93%, and 52%. Thus, a longer life span is possible with adequate supportive care. Advances in respiratory care have resulted in increased the survival of patients with severe disease.[2]

Most SMA type II patients live to the third decade, and many could live to the fourth and fifth decade. In this patient age, the survival rate is approximately 52% through 40 years old with adequate support, especially respiratory management. The most deadly complication is from respiratory complication, which includes pneumonia, aspiration, mucus retention, respiratory dysfunction until failure. Respiratory complications are more likely due to airway clearance problem and weak cough ability tend to have recurrent pneumonia, resulting in recurrent hospitalization.

Spinal muscular atrophy II disease onset is usually more insidious than SMA type I. Generalized hypotonia, symmetrical weakness, and delayed motor milestones are hallmarks of SMA II. Weakness usually involves proximal muscle more than distal muscle and lower extremity more than upper. Muscle wasting tends to be more clearly visible in SMA II. Deep tendon reflexes are depressed and usually absent in lower extremities. Progressive kyphoscoliosis and neuromuscular restrictive lung disease are almost invariably seen in the late first decade due to intercostal muscle weakness. Contractures of hip flexors, tensor fascia lata, hamstrings, triceps surae, and elbow and finger flexors are common. Hypotonic hip dislocation has been noted in type II patients. Cognition of these patients is normal and verbal intelligence may be above average.[1]

Adult-onset SMA and SMA III patients can live average life spans with a relatively benign disease course. Furthermore, with the rapid advancement of rehabilitation technology, many SMA II patients live well into adulthood, and success has been reported in this population.[3] ADL in this patient is full dependency and will always need help from a caregiver. But productivity could be optimized with help from an assistive device, given this patient has above average cognitive ability.

Patients with SMA could be experience difficulty during feedings, which can lead to aspiration with recurrent respiratory infection. It is also proposed that patients with SMA have a high incidence of silent gastroesophageal reflux disease (GERD), contributing to aspiration. Patients with SMA are at risk of constipation that could worsen reflux or even respiratory symptoms if severe enough.[4]

Muscle weakness limits the motoric function of trunk, upper and lower extremities resulting in contracture formation, spinal deformity, limited mobility and activities of daily living (ADL). Fracture and hip subluxation

are commonly seen in milder type II and type III SMA patients. Kyphoscoliosis develops in more than 50% children with SMA, mostly in non-ambulatory children or in those who lose the ability to walk.[4]

Respiratory failure is the primary cause of mortality in patients at the more severe end of the disease spectrum, namely type I and II SMA. Weak intercostal muscle with relatively preserved diaphragm strength resulting in a bell-shaped chest, pectus excavatum, and in some cases, underdevelopment of lungs. Type II SMA have weak intercostal muscles with scoliosis contributing to progressive restrictive lung disease. This restrictive lung disease results in the insidious onset of sleep hypoventilation. Patients with SMA and respiratory muscle weakness also have a weak cough which impairs airway clearance. The risk of hypoxemia from mucus plugging, especially during acute illness and recurrent infection, is high. It is recommended that patients at risk of mucus plugging be monitored with overnight oximetry during acute illness, and an assisted airway clearance methods should be employed.[5] Patient's SpO2 was between 95-98%. Patient did not experience sleep disturbance, but SpO2 diary should be done due to monitor hypoventilation, especially at night.

A minimum of 160 L/m Peak Cough Flow (PCF) is used to measure cough effectiveness, and this is the best indicator for tracheostomy tube removal irrespective of remaining pulmonary function. Patients with Vital Capacity (VC) less than 1,500 mL will have assisted PCF measured from a maximally stacked volume of air and an abdominal thrust delivered simultaneously with the glottic opening. Assisted PCF can be significantly increased in patients receiving maximal insufflations followed by manual thrusts for assisted coughing. Techniques of manually assisted coughing involve different hand and arm placements for expiratory cycle thrusts. An epigastric thrust with one hand while applying counterpressure across the chest to avoid paradoxical chest expansion with the other arm further increases assisted PCF for 20% of patients. Abdominal compressions should not be used in 1 to 1.5 hours following a meal. When those were inadequate, the most effective alternative for generating optimal PCF and clearing airway secretions is the mechanical insufflation-exsufflation (M I-E).[6] In this patient, PCF was still good. Manually assisted coughing could be done by his caregiver, with manual ambu-bag as air stacking procedure followed with abdominal thrust.

Patient with respiratory muscle weakness will need the air stacking procedure which is considered as a respiratory orthosis to increase ventilation starting from overnight (nocturnal) noninvasive ventilation device to fulltime support (daytime and overnight NIV support). Frequent blood gas analysis is needed to monitor CO_2 level and hypoxemia.[1] Hypercapnia symptoms in this patient were unspecific, such as fatigue, feeling dypsnea, shortness of breath. Patient could not afford any objective monitoring with device like capnograph, therefore measurement could only be done when patient come to the hospital. The patient has fingertip pulse oximetry which can show the current peripheral oxygen saturation.

Many of these patients experience ventilation insufficiency and require ventilator. Respiratory muscle dysfunction amenable to treatment by respiratory muscle aids occurs in many people with the SMA diagnosis. Surveys in the United States, Western Europe, and Japan indicate that the use of home mechanical ventilation is increasing rapidly.[7]

In this patient, there were no contraindication for NIV. Patient's SpO2 was still above 95%, had an adequate caregiver support, and had good cognitive function. The patient could only use nasal prong due to lip sealing problem. His daily caregiver was not professional medical provider but already well-trained about the procedure of applying NIV setting and how to do cardiopulmonary resuscitaton (CPR) if in an emergency situation.

Conclusion

Respiratory condition in adult neglected Spinal Muscular Atrophy Type II patients need a well-planned routine monitoring from physical medicine and rehabilitation team and the caregiver. Respiratory rehabilitation comprehensive management can optimize the patient's performance and prevention of pulmonary infection or further complication so patient can do daily activity regularly and increase his quality of life.

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Conflicts of Interest

The authors declare that there are no conflicts of interest.

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