

# A Case of Spinal and Bulbar Muscular Atrophy Requiring Amputation and Airway Support

Haruka Makinae<sup>1\*</sup>, Taichiro Takamatsu<sup>2</sup>, Akimasa Suzuki<sup>2</sup> and Hidenobu Kamohara<sup>1</sup>

<sup>1</sup>Division of Critical Care Medicine, Tokyo Medical University Hachioji Medical Center, Hachioji, Tokyo, Japan

<sup>2</sup>Department of Orthopaedics, Tokyo Medical University Hachioji Medical Center, Hachioji, Tokyo, Japan

## Abstract

Spinal and bulbar muscular atrophy (SBMA) is a rare neuromuscular disorder. Here, we present the case of a 53-year-old male diagnosed with SBMA, who underwent left leg amputation for a compound fracture with gas gangrene. The patient was hospitalized with gas gangrene and sepsis, and required non-invasive positive pressure ventilation, inotropes, and continuous hemodiafiltration. The patient gave consent for mechanical ventilation and underwent an amputation. Before and after the amputation, the patient had recurrent severe atelectasis of the left lower lobe, and large emboli composed of coagulated blood cells and sputum were extracted. SBMA itself is not curable, although the morbidity and complications related to SBMA can be managed using essential procedures.

**Keywords:** Atelectasis • Gas gangrene • Spinal and bulbar muscular atrophy • X-linked disease • Non-occlusive mesenteric ischemia

**List of Abbreviations:** PEF- Peak expiratory flow; SBMA- Spinal and bulbar muscular atrophy

## Introduction

Spinal and bulbar muscular atrophy (SBMA) is an X-linked recessive disease caused by mutations in androgen receptor genes. This mutation is only found one in 40,000 individuals and is only expressed in males. The detection of CAG repeat expansion in the androgen receptor gene on the X chromosome is the key to diagnosis. Disease progression is slow and is accompanied by a decline in muscle strength [1-3], which can lead to complex multi-organ involvement. The non-neurological features of SBMA include gynecomastia, insulin resistance, metabolic syndrome with increased body mass index, elevated serum cholesterol and triglyceride levels, testicular atrophy, and reduced fertility [4-6]. Here, we present the case of a 53-year-old male diagnosed with SBMA who underwent left leg amputation for a compound fracture with gas gangrene. This article was presented at the 50th Annual Meeting of the Japanese Society of Intensive Care Medicine on March 4, 2023.

## Case Presentation

The 53-year-old male in this case had been diagnosed with diabetes mellitus 20 years previously and had undergone dialysis for diabetic renal failure. The patient had also suffered a left fibula fracture 15 years prior but refused surgical procedures because he was afraid that he would require mechanical ventilation for the rest of his life. The patient's daily activity level was limited because he required a wheelchair owing to muscle atrophy. An expansion of the CAG repeat was detected in the androgen receptor gene,

**\*Address for Correspondence:** Haruka Makinae, Division of Critical Care Medicine, Tokyo Medical University Hachioji Medical Center, Hachioji, Tokyo, Japan, Tel: +81-42-665-5611, E-mail: cyp02060@nifty.com

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**Received:** 01 February, 2023; Manuscript No. jccr-23-92511; **Editor Assigned:** 03 February, 2023; PreQC No. P-92511; **Reviewed:** 14 February, 2023; QC No. Q-92511; **Revised:** 20 February, 2023, Manuscript No. R-92511; **Published:** 27 February, 2023, DOI: 10.37421/2165-7920.2023.13.1554

and the patient was diagnosed with SBMA. In this case, the patient fell from his wheelchair and was diagnosed with a left tibial fracture. He was hospitalized for gas gangrene and sepsis, and required non-invasive positive pressure ventilation, inotropes, and continuous hemodiafiltration.

The patient gave consent for mechanical ventilation and underwent an urgent amputation. Before and after the amputation, the patient had recurrent severe atelectasis of the left lower lobe, and large emboli composed of coagulated blood cells and sputum were extracted. The patient experienced repeated airway complications; however, the frequency decreased over time. The patient was discharged from the intensive care unit on the 8th postoperative day. Although he had started rehabilitation to perform his daily living activities in a wheelchair, on the 14th postoperative day, he suddenly developed symptoms of non-occlusive mesenteric ischemia and died the following day.

## Discussion

Evans R, et al. reported a surgical case involving a patient with SBMA who was successfully extubated; however, the patient was able to walk approximately 100 m at the time of surgery [1]. In the present case, the patient was already in a wheelchair as the disease advanced. Wang AY, et al. suggested that pulmonary complications could be a major cause of mortality, especially in patients with higher level spinal cord injury [7]. They also found a direct relationship between motor level and peak expiratory flow (PEF) [7]. Yamada S, et al. compared the characteristics of SBMA and amyotrophic lateral sclerosis and reported that the predicted values of PEF and forced vital capacity were lower in patients with SBMA, although only the PEF value was significantly lower than that in patients with amyotrophic lateral sclerosis [8]. The authors mentioned that PEF is generated by explosive muscle power during effort-dependent procedures. In our case, recurrent airway obstruction could have been caused by a reduction in the PEF, which is the driving force for coughing. We performed repeated airway suction via bronchoscopy and extracted large emboli from the lower lobes. Emboli may have formed by the accumulation of uncleared sputum. After the extraction of the large emboli, the

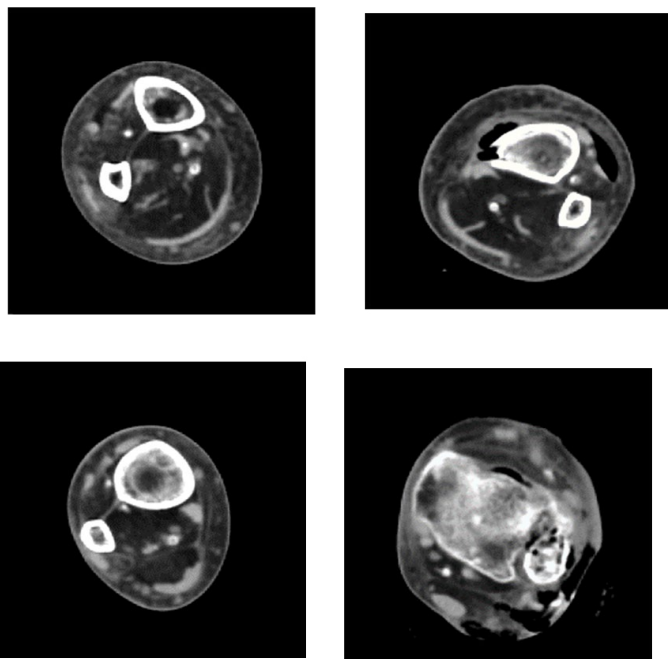


Figure 1. Preoperative CT scan showing gas gangrene of the left foot.

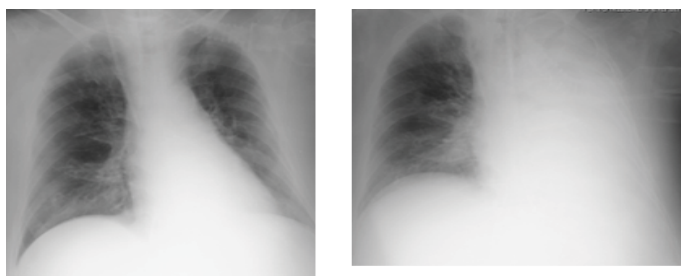


Figure 2. Postoperative chest X ray.

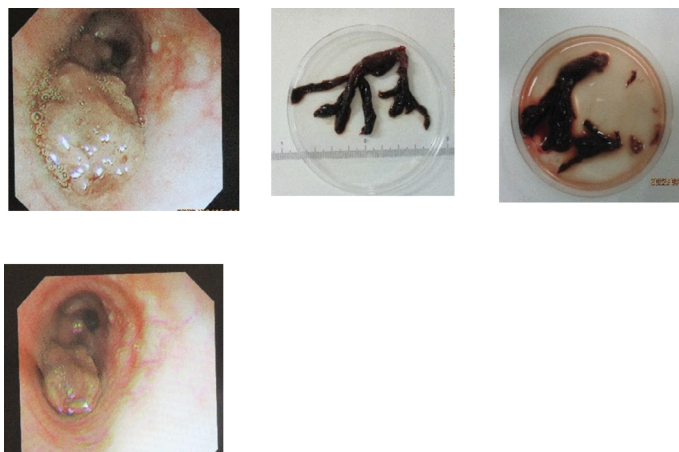


Figure 3. Postoperative endoscope of trachea.

patient was discharged from the intensive care unit, although he still required airway suction several times daily (Figures 1-3).

## Conclusion

SBMA is an incurable disease that progresses slowly. Mechanical ventilation and support for daily activities are essential lifelong requirements. In this case, we were able to perform airway management and deliver the required treatment, including a surgical procedure under general anesthesia

## Acknowledgements

This study was partially supported by a Grant-in-Aid for Scientific Research C (22K09173) from the Japan Society for the Promotion of Science (Hidenobu Kamohara).

## Disclosures

All authors have no conflicts of interest to declare. The patient in the present case suffered a muscular disease and died of non-occlusive mesenteric ischemia; thus, written consent was signed by the patient's brother. This study was conducted according to the principles of the Declaration of Helsinki. Ethical review board approval was not required for this study.

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**How to cite this article:** Makinae, Haruka, Taichiro Takamatsu, Akimasa Suzuki and Hidenobu Kamohara. "A Case of Spinal and Bulbar Muscular Atrophy Requiring Amputation and Airway Support." *Clin Case Rep* 13 (2023): 1554.