

# Fatal Case of Dysphagia Induced by Neurofibromatosis Type II

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**Abstract** A 17-year-old boy was found in an unconscious state. He had had neurofibromatosis type 2 (NF2) since 9 years old. His history included multiple operations for NF2 at the left jugular vein foramen, thoracic spinal cord, and cervical spinal cord at 9 and 10 years old. However, remaining bilateral acoustic tumors resulted in the onset of aspirated pneumonia within a few months. He was found unconscious in his dormitory. When emergency medical technicians checked him, he was in cardiac arrest. Saburra was identified in his mouth. He did not achieve return of spontaneous circulation with advanced cardiac life support. Autopsy imaging revealed residual bilateral acoustic tumors compressing the brain stem. NF2 is a serious disease that can rapidly become life-threatening. We hope new treatments that will improve the survival of NF2 patients will be developed soon.

**Keywords:** neurofibromatosis, fatality, dysphagia

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## 1. Introduction

Neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2) are tumor-suppressor syndromes. Systematic investigations of the pathways impacted by the loss of the function of neurofibromin (encoded by NF1) and merlin (encoded by NF2) have led to advances in treatments for patients with NF1 and NF2. [1]

The manifestations of NF1 are as a neurocutaneous syndrome, with hallmark lesions involving the skin, including café au lait spots, intertriginous freckling, and cutaneous and subcutaneous neurofibromas. [1] The hallmark lesions of NF2 are bilateral vestibular schwannomas, which are the most common cause of morbidity among patients with NF2, resulting in bilateral sensorineural hearing loss, tinnitus, difficulty maintaining balance, and ultimately deafness, facial nerve weakness, and possible brainstem compression. [1]

We herein report a fatal case of NF2 in a young patient.

## 2. Case Presentation

A 17-year-old boy was found in an unconscious state. He had NF2 since 9 years old. His history included multiple operations for NF2 at the left jugular vein foramen, thoracic spinal cord, and cervical spinal cord at 9 and 10 years old. However, remaining bilateral acoustic tumors resulted in the onset of aspirated pneumonia within

a few months. He was found at unconsciousness state in a toilet at a diet room in a dormitory. When emergency medical technicians checked him, he was in a cardiac arrest. Initial rhythm was asystole. Saburra was identified in his mouth, and after its resolution, he underwent tracheal intubation and advanced cardiac life support.

A venous blood gas analysis on arrival at out hospital revealed the following: pH, 6.657; PaCO<sub>2</sub>, 156; HCO<sub>3</sub><sup>-</sup>, 16.4 mmol/L, base excess -42.5 mmol/L, glucose, 564 mg/dL and potassium, 11.0 mEq/L. He did not achieve return of spontaneous circulation with advanced cardiac life support. Autopsy imaging revealed residual bilateral acoustic tumors compressing the brain stem (Figure 1).



**Figure 1. Autopsy imaging.** The autopsy imaging revealed residual bilateral acoustic tumors (arrow) compressing the brain stem

### 3. Discussion

Aboukais et al. performed a retrospective review of 80 patients with NF2 disease and investigated the cause of death.[2] Among them, there were seven deaths. The mean age at the diagnosis of the patients who died was 26 years old, and the mean age of death was 38.9 years old (youngest age: 25 years old). The present case was even younger than those cases, but it was Wu et al. who reported the youngest fatal cases (9 years old) due to brain metastasis of a malignant peripheral nerve sheath tumor in an NF2 patient. [3] The causes of death in Aboukais et al. were suicide in one patient, hematoma after surgical removal of grade IV vestibular schwannoma in one patient, aspiration pneumonia after swallowing issues in three patients, intracranial hypertension related to growth of multiple meningiomas in one patient, and brachial plexus sarcoma grade 3 in one patient. [3] Based on these findings, the authors concluded that NF2 is a serious disease that can rapidly become life-threatening.

The presence of lower cranial nerves schwannomas is a poor prognostic factor, similar to the present patient. Aspirated pneumonia due to dysphagia is associated with poor short- and long-term prognoses, even in children. [4,5] NF experts and patient representatives support the prioritization of the development of drug trials for improving the prognosis of patients with NF2. [6]

### 4. Conclusion

NF2 is a serious disease that can rapidly become life-threatening. We hope new treatments that will

improve the survival of NF2 patients will be developed soon.

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