A rare challenge in general surgery: double surgical procedure for large and small bowel obstruction in a patient with Gerstmann-Sträussler-Scheinker syndrome

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SUMMARY Gerstmann-Sträussler-Scheinker syndrome (GSS) is a rare, infectious syndrome related to a mutation in the prion protein gene. Described here are the challenges posed by surgery for a patient with GSS. A 61-yr-old woman with GSS was admitted to this department and underwent surgery twice for large and small bowel obstruction. This is the first report of two major surgical procedures in a patient with GSS. Experiences with this case and precautions when using a disposable device during endotracheal intubation and a surgical procedure to manage a patient with GSS are described.

Keywords Gerstmann-Sträussler-Scheinker syndrome, general surgery, bowel obstruction

Gerstmann-Sträussler-Scheinker Syndrome (GSS) is a rare prion disease (PD). This familial (autosomal dominant), fatal neurodegenerative disease affects patients from 20 to 60 years of age and is related to a prion protein (PrP) mutation in the group of subacute spongiform encephalopathies (SEs) (1). Human SEs include Creutzfeldt-Jakob disease, GSS, Kuru, and fatal familial insomnia. GSS is characterized by amyloid deposition in the cerebral parenchyma or blood vessels (2). Symptoms include dysarthria, progressive cerebellar truncal ataxia, pyramidal signs, and adult-onset dementia. Some studies have suggested that a PD might be transmitted by blood or plasma-derived products from patients during the prodromal stage. In animal studies, intracerebral inoculation of infected cells has been associated with development of disease, and infectivity was also detected in the blood (3). Given the lack of information, the resistance of PrP to conventional sterilizing measures is a major problem. Current recommendations are to identify at-risk patients and to use disposable devices during endotracheal intubation, spinal anesthesia, and surgery (4).

A 61-yr-old woman was admitted to this Hospital for abdominal pain. At the age of 53, she complained of dysarthria, spastic paraparesis, cognitive decline, amyotrophy, depression, cerebellar ataxia, pseudo-bulbar palsy, and absent tendon reflexes in the lower limbs. Her father and sister had the same symptoms. A typical mutation of the 102nd amino acid (PRNP-p.D202N) and a drastic change were found in the normal prion gene. She was then diagnosed with classical GSS. Her Unified Parkinson's Disease Rating Scale III (UPDRSIII) score was 26 and her Hoehn and Yahr scale score was 3 (5). The patient was admitted to the emergency department for abdominal pain. Laboratory results revealed an increased white blood cell count (WBC) [14 × 10⁹/L (NV 4–10 × 10⁹)] and elevated C-reactive protein (CRP) level [33.3 mg/L (NV < 1.0)]. A CT scan of the abdomen and thorax revealed sigmoid volvulus. The endoscopic evaluation confirmed sigmoid stenosis and mucosal necrosis. After an interview with the patient's caregivers who are actively present in the patient's life and fully dedicated to her medical and physical care and a consultation with anesthesiologists and physiotherapists, a plan was formulated to use general anesthesia with tracheal intubation in order to perform an explorative laparotomy.

A volvulus of the dolichosigmoid colon with bowel obstruction and ischemia was detected intraoperatively. In order to avoid anastomotic complications, the patient underwent a Hartmann's resection with end-colostomy. The highest level of protection was used during surgery. In addition to disposable masks and caps, all medical staff wore gowns and gloves. Disposable surgical instruments and anesthetic equipment were used. The scrub team was equipped with full personal protective equipment. The number of surgical, nursing, and anesthesia team members was limited to

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the minimum required to perform the surgery. After confirming full recovery of muscle strength, double-burst stimulation, and spontaneous eye opening, the tracheal tube was removed with no incidence of airway obstruction. She was admitted to the intensive care unit on day 1 postoperatively, where her vital signs were stable. She was extubated on day 1 postoperatively and a percutaneous mini-tracheostomy for pulmonary aspiration was performed. The postoperative course was uneventful. The patient underwent physiotherapy sessions and was discharged on day 6 postoperatively without complications.

One year later, the patient was seen by the emergency department for small bowel occlusion. Blood tests revealed increased CRP (9 mg/L (NV < 1.0)), WBC (12 × 10⁹/L (NV 4.0-9.0)), and plasma lactate (3.9 mmol/L). Neurological symptoms were stable and the level of home care was optimal. A second emergency laparotomy was performed, and small bowel ischemia due to ileal obstruction was detected intraoperatively. A small bowel resection was performed and an endoscopic gastrostomy (PEG) tube was placed for enteral nutrition. The same level of protection as in the first surgical procedure was used. The postoperative course was uneventful, and the patient was discharged on day 9 postoperatively with a permanent urinary catheter and PEG tube for enteral nutrition (Table 1).

There are no cases of a patient with GSS undergoing multiple abdominal surgeries under general anesthesia in the literature (according to a search for relevant articles on PubMed and Embase using the terms “GSS” OR “PD” AND “Surgery”).

The challenge in the surgical treatment of patients with GSS involves intra- and post-operative anesthesiological risks (e.g., airway obstruction, bronchospasm, or pneumonia), surgical risks (e.g., immobility, dysphagia, or impaired canalization), and risks of infection. PrP is present in the central nervous system, appendix, and lymphatic tissues and is resistant to inactivation by radiation, heat, or aggressive chemical treatments. Patients with GSS must be managed with specific precautions to prevent infections.

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### Table 1. Patient characteristics

| Age (yrs) | Neurological Symptoms | Abdominal Symptoms | WBC (× 10⁹/L) | CRP (mg/L) | Radiological Findings | Surgical Procedure | Anesthetic Outcome | Surgical Outcome |
|-----------|-----------------------|--------------------|---------------|------------|-----------------------|-------------------|-------------------|-----------------|
| 1st surgery | 61 yrs | Dysearthria; Spastic Paraparesis; Dementia; Cerebellar Ataxia; Pseudo-bulbar Palsy; Tendon Reflexes Absent | Abdominal Pain; Nausea and Vomiting; Bowel Obstruction | 14 | 33 | Colonic Distension; Bowel Obstruction; Sigmoid Volvulus; No Signs of Perforation | Laparoscopic Hartmann's Resection with end Colostomy | No Airway Obstruction; Extubated on Day 1 | Uneventful; Discharged on Day 6 Postop |
| 2nd surgery | 62 yrs | Stable | Abdominal Pain; Nausea and Vomiting; Small Bowel Obstruction | 12 | 9 | Bowel Distention; Closed-loop Obstruction | Laparotomic Small Bowel Resection; PEG Tube Placement | Uneventful | Uneventful; Discharged on Day 9 Postop |

yr: years; WBC: white blood cell count; CRP: C-reactive protein.
References

1. Boellaard JW, Schlote W. Subakute spongiforme Encephalopathie mit multiformer Plaquebildung. "Eigenartige familiär-hereditäre Krankheit des Zentralnervensystems [spino-cerebellare Atrophie mit Demenz, Plaques und plaqueähnlichen Ablagerungen im Klein- und Grosshirn" (Gerstmann, Sträussler, Scheinker)] [Subacute spongiform encephalopathy with multiform plaque formation. "Peculiar familial-hereditary disease of CNS [spinocerebellar atrophy with dementia, plaques, and plaque-like deposits in cerebellum and cerebrum" (Gerstmann, Sträussler, Scheinker)] [author's transl]]. Acta Neuropathol. 1980; 49:205-212. (in German)

2. Ghetti B, Piccardo P, Zanusso G. Dominantly inherited prion protein cerebral amyloidoses – A modern view of Gerstmann-Sträussler-Scheinker. Handb Clin Neurol. 2018; 153:243-269.

3. Geschwind MD. Prion Diseases. Continuum (Minneap Minn). 2015; 21 (6 Neuroinfectious Disease): 1612-1638.

4. Nakamura M, Ogata M, Matsuo Y, Sata T. Anesthetic management of a patient with Gerstmann-Sträussler-Scheinker syndrome (mutation of prion protein). Anesth Analg. 2006; 102:1285-1286.

5. Leng B, Sun H, Zhao J, Liu Y, Shen T, Liu W, Liu X, Tan M, Li F, Zhang J, Li Z. Plasma exosomal prion protein levels are correlated with cognitive decline in PD patients. Neurosci Lett. 2020; 723:134866.

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