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A case report: Anaesthetic management in a child with Tay-Sachs disease undergoing feeding gastrostomy

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Abstract
Tay-Sachs disease (TSD) is a rare autosomal recessive genetic disorder caused by a deficiency of the enzyme hexosaminidase-A. Infants with TSD typically develop normally for the first few months of life, after which they experience progressive loss of motor functions, cognitive decline, seizures, blindness and difficulty in swallowing. These complications often lead to death by the age of three to five years. A two-year-old boy with TSD presented with swallowing difficulties, choking episodes, and intermittent seizures over the preceding two months. He was scheduled for a feeding gastrostomy to address his nutritional needs. Pre-anaesthetic evaluation revealed severe disease manifestations, including profound cognitive impairment, unresponsiveness to painful stimuli, blindness, hearing loss, and ongoing convulsions. His medication regimen included clobazam, levetiracetam, baclofen, perampanel, and glycopyrrolate, administered via a nasogastric tube. During the preoperative period, the patient experienced convulsions, which were managed with intravenous midazolam. General anaesthesia, rapid sequence induction with glycopyrrolate, fentanyl, propofol, and rocuronium, followed by endotracheal intubation was done. Maintenance anaesthesia included oxygen, nitrous oxide, sevoflurane and rocuronium. Neuromuscular monitoring was done throughout the procedure. After surgery, neuromuscular blockade was reversed with neostigmine, and the patient was extubated upon demonstrating adequate respiratory effort.
This case highlights the importance of thorough pre-anaesthetic assessment and careful intraoperative management in patients with TSD undergoing surgical procedures.

Keywords: Infantile Tay Sachs disease, Hexa gene A mutation, Neurodegenerative disorder, anesthetic management

Introduction
Tay-Sachs Disease (TSD) is a rare, autosomal recessive neurodegenerative disorder caused by mutations in the HEXA gene, leading to a deficiency of the enzyme beta-hexosaminidase A. This results in the accumulation of GM2 gangliosides in neurons, causing progressive neurological deterioration. Infantile Tay-Sachs disease (ITSD), the most severe form, manifests with developmental regression, hypotonia, seizures, and respiratory compromise. The anaesthetic management of these patients is complex due to their multisystem involvement and heightened sensitivity to various anaesthetic agents.
We report a successful anesthetic management of a case of Infantile Tay-Sachs disease posted for feeding gastrostomy.

Case report: A two-year-old boy, weighing 12 kg, known to have Tay-Sachs disease since six months of age, was admitted to the hospital with difficulty in swallowing, choking episodes, and intermittent seizures since two months. The child was scheduled for a feeding gastrostomy surgery. A thorough pre-anaesthetic evaluation was conducted, a day before the surgery after obtaining informed parental consent. The assessment revealed a severe form of Tay-Sachs disease, with the child exhibiting: Severely impaired higher mental functions, with no response to painful stimuli, Blindness and hearing loss, Difficulty in swallowing with frequent choking episodes and Recurrent convulsions. The child was on the following medications, administered via a nasogastric tube: Syrup Clobazam 5 mg BD, Syrup Levetiracetam 100 mg BD, Syrup Baclofen 2.5 mg BD, Syrup Perampanel 4 mg HS, Tablet Glycopyrrolate 1 mg BD. Patient RT feed was withheld and 8 hours of fasting was ensured on the day of surgery. 0.45% DNS was started and RBS checked 2nd hourly to prevent

hypoglycaemia. Patient received injection ondansetron 0.1mg /kg IV and injection Ranitidine 1.5 mg/kg IV.

General anaesthesia with endotracheal intubation was planned. A difficult airway cart was kept ready. The patient was shifted to the operating room and standard monitors were applied (pulse oximetry, non-invasive blood pressure, ECG). The oxygen saturation values on

admission to the operating room were 100% with Blood Pressure of 90/60 mmhg and pulse 122/min was recorded. On-table, before induction, the patient experienced seizures, which was managed with intravenous Midazolam. General anaesthesia was administered by rapid sequence induction with Injection Glycopyrrolate 0.06 mg, Injection fentanyl 25 mcg, Injection Propofol 25 mg and Injection rocuronium 12 mg. Intubated with 4.0 mm ID cuffed endotracheal tube. Anaesthesia was maintained with Oxygen, Nitrous Oxide, Sevoflurane and Intermittent top ups of Injection Rocuronium given. Throughout the procedure neuromuscular monitoring was done to facilitate administration of neuromuscular blocking agents as well as at the time of extubation. Once the procedure was over Neuromuscular blockade was reversed with Injection Neostigmine 0.6 mg. Trachea was successfully extubated after through oropharyngeal suctioning and confirming adequate respiratory efforts and the generation of adequate tidal volume. Post extubation vitals were stable, patient maintained room air oxygen saturation of 99%. Patient received injection paracetamol 15mg/kg IV for post operative analgesia. patient shifted to recovery room. The child was closely monitored postoperatively for seizures, respiratory distress and aspiration.

Discussion

The infantile form of Tay Sachs disease is the most common and severe type, typically presenting between 3 to 6 months of age. Following initial normal development, the infant develops hypotonia, blindness, progressive hearing loss, seizures, swallowing difficulty as the disease progress [1, 2]. On fundoscopic examination a cherry red spot in retina seen is the hallmark finding of TSD. Macrocephaly also develops due to swelling of the brain due to GM2 accumulation. Our patient presented with almost all the signs of severe form of the disease. Our child also had macrocephaly, hence difficult intubation cart was kept ready [3, 4]. In view of history of choking episodes, feeding gastrostomy was planned to meet the nutritional needs of the patient. Extensive literature search revealed only 2-3 case reports exists regarding anaesthetic management of Tay Sachs Disease [5]. In view of recurrent choking episodes and the presence of Ryles Tube, a rapid sequence induction was planned. Patient received all his medications on the day of surgery through Ryles Tube. We avoided all the drugs which were known to be epileptogenic in view of patient's seizure history. Even then patient threw seizure on table which was controlled with intravenous midazolam. Due to hyperkalaemia risk, succinylcholine was avoided. Injection Rocuronium was used to facilitate Rapid Sequence Induction intubation in the dose of 1mg/kg. Neuromuscular monitoring were used to guide the use of muscle relaxant as well as for the extubation.

Although Tay Sachs Disease primarily affects the nervous system and cardiac involvement is rare, it can cause autonomic dysfunction leads to haemodynamic instability [5, 6]. However, our patient did not show any evidence of autonomic dysfunction. Haemodynamic parameters remained stable intraoperatively. In view of feeding difficulties, TSD patients are at a risk of hypoglycaemia and electrolyte abnormalities. In our patient pre operative electrolytes and sugars were normal. We also monitored RBS intra operatively and it was maintained within normal values. Restrictive lung disease may also be present in these patients secondary to the involvement of the diaphragm [7, 8, 9]. Keeping this in mind patient was mechanically ventilated with pressure controlled ventilation to prevent barotrauma. Our patient was completely unresponsive to any visual or auditory communication, hence extubation was done with TOF monitoring and once adequate respiratory effort was achieved. Patient was monitored carefully in PACU in view of risk of regurgitation and aspiration. As our patient was unresponsive, analgesics were administered in a time bound manner keeping the haemodynamic parameters in mind.

Conclusion

Infantile Tay-Sachs disease is a devastating genetic disorder with no cure, leading to severe neurodegeneration and early mortality. Early diagnosis through enzyme assays and genetic testing is crucial for genetic counselling and family planning. Research into gene therapy and enzyme replacement therapy is ongoing, offering hope for potential future treatments.

The anaesthetic management of infants with Tay-Sachs disease requires careful planning and multidisciplinary collaboration due to their complex physiological impairments. A tailored approach considering respiratory compromise, airway management, and aspiration risk is essential to ensure safe perioperative care.

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