Anaesthetic management of caesarean section in a patient with Pompe disease

Sir,

Pompe disease is an autosomal recessive type II glycogen storage disorder affecting 1 in 40,000 live births.It is caused by the deficiency of lysosomal acid α-glucosidase (GAA) enzyme that degrades lysosomal bound glycogen to glucose.[1] We present the anaesthetic management of a 21-year-old primigravida at 37 weeks of gestation, a known case of juvenile Pompe's disease, posted for elective caeserean section. Her symptoms started at 10 years when she noticed difficulty in getting up from squatting position. The diagnosis was confirmed by leukocyte enzyme analysis and muscle biopsy. She was started on enzyme replacement therapy (ERT) alglucosidase alfa 20mg/kg intravenous infusion over 5h every 2 weeks.

She conceived spontaneously and continued to get ERT throughout her pregnancy. At 34 weeks gestation, she was admitted for safe confinement and was started on oral ubiquinone 90mg once daily and injection enoxaparin 0.4 mg subcutaneously once daily. Blood investigations and echocardiogram were normal. Pulmonary function tests in standing and sitting position showed severe restriction [Table 1 and Figure 1].

For caeserean section, subarachnoid block was given. She delivered a live healthy female baby of 2.8 kg. After delivery, oxytocin infusion was started. The procedure was uneventful. Postoperative analgesia was provided with ultrasound-guided bilateral transversus abdominis plane block with 30 ml of 0.1% ropivacaine and intravenous 1g paracetamol 8th hourly. She had an uneventful postoperative period and received one more dose of ERT infusion. She was discharged on the 5th postoperative day.

Based on the age of onset, Pompe disease is classified into infantile and late-onset. Infantile form is severe and presents with failure to thrive and feeding difficulties. Late-onset Pompe disease can present at any age. They can also have kyphoscoliosis.[2] Proximal muscles of the lower limb and paraspinal trunk muscles are affected initially followed by diaphragm. Pregnancy is risky in these patients as the mass effect of the uterus could further compromise the respiratory function.Elective caeserean section is usually preferred in these patients.

Enzyme replacement therapy (ERT) with alglucosidase alfa, though expensive is found to be effective and safe

Table 1: Pulmonary function test in erect and supine position

	Supine PFT		Erect PFT	
	Volume	% Predicted	Volume	% Predicted
FVC (L)	1.31	44	1.38	49
FEVI (L)	1.21	50	1.25	51
FEVI Ratio	0.92	110	0.91	108
PEF (L/min)	190	48	199	50
FEF25-75 (L/s)	1.95	49	1.89	47

PFT-Pulmonary function tests; FVC-Forced vital capacity; FEV1-Forced expiratory volume measured during first second; PEF-Peak expiratory flow; FEF-Forced expiratory flow

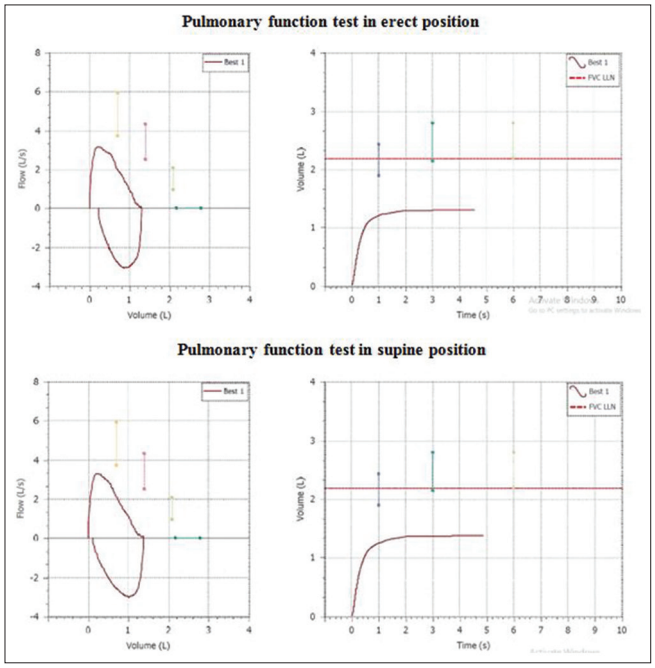

Figure 1: Pulmonary function tests

during pregnancy and lactation.[3] It is secreted into the breast milk but returns to the pre-infusion level after 24h. Hence, lactating mothers are advised against breastfeeding for 24h after receiving ERT.[4] They are prone to deep venous thrombosis because of restricted mobility, hence started on heparin preoperatively. They are advised to take high protein diet to avoid muscle wasting. Ubiquinone and L-carnitine are supplemented to increase fat metabolism and decrease protein degradation. Ephedrine is used to stimulate glycogen degradation.[1]

Preoperative assessment of respiratory function by comparing erect and supine vital capacities will help in assessing diaphragmatic weakness. A drop in functional vital capacity of ≥10% from upright to supine position is suggestive of diaphragmatic weakness. Arterial blood gas is done in severe cases to rule out carbon dioxide retention and hypoxia. Due to restrictive respiratory functions, regional anaesthetic technique is preferred over general anaesthesia.[1] Epidural or combined spinal–epidural anaesthesia could be used for caeserean section.[5,6] But this could be difficult if there is associated scoliosis. The cough, gag, and swallowing reflexes are affected increasing the risk of aspiration. Hence if general anaesthesia is required, airway has to be protected by endotracheal intubation. Difficult intubation has to be anticipated as macroglossia might make visualisation of the vocal cords difficult. Suxamethonium is contraindicated as it can cause severe hyperkalemia. Non-depolarising neuromuscular blockers are better avoided or if required reduced doses are recommended. Respiratory and myocardial depressant drugs have to be cautiously used. Use of systemic opioids should be restricted to avoid respiratory complications. Postoperative pain has to be managed with epidural analgesia or nerve blocks.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and othe r clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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