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Case Report

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Case of Wilson's Disease with Fracture Femur–An Anaesthetic Challenge

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ABSTRACT

Background: Wilson's disease is an inherited disorder of altered copper metabolism characterized by hepatic, renal, ocular and haematological dysfunction. It is a rare genetic disease and very few cases are reported on anaesthetic management of these cases.

Methods: We present anaesthetic management of a case of an 18-year-old female with Wilson's disease posted for open reduction and internal fixation for sub-trochanteric femur fracture. Our anaesthesia of choice was combined spinal epidural anaesthesia.

Results: The case was successfully managed under a combined spinal epidural with post-op ICU monitoring.

Conclusion: Anaesthetic management of a patient with Wilson's disease requires a multidisciplinary approach utilizing the expertise of an anaesthesiologist, physician, orthopaedician, neurologist and detailed knowledge of the organ systems involved.

Key-words: Combined spinal epidural, Dexmedetomidine induced Apnea, Kayser Fleischer Rings, Wilsons disease

INTRODUCTION

Wilson's disease is a rare autosomal recessive genetic disorder characterized by impaired copper metabolism. The incidence of Wilson's disease is 0.5 in 1 lakh [1]. In Wilsons disease, excessive amounts of free copper are deposited in the liver, kidney, brain and cause oxidative damage in these organs. These patients present with challenges like deranged hepatic and renal function, hemodynamic instability, bulbar involvement, and respiratory insufficiency.

This case report presents the perioperative management of a case of Wilson's disease with sub-trochanteric femur fracture with unique challenges peri-operatively due to disease pathology involving multiple organs.

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CASE REPORT

An 18-year-old female born of non-consanguineous marriage, weighing 30 kgs diagnosed with Wilson's disease was admitted with sub-trochanteric femur fracture of the right side for open reduction and internal fixation.

Four years back, she was alright when she started complaining of a change in voice, slurring of speech, and imbalance while walking.

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Her complaints got aggravated to difficulty in breathing, swallowing and weakness in bilateral upper and lower limbs. She became irritable and stopped interacting with relatives. On examination, she had dysdiadochokinesia, short stepping gait, decreased planter reflex, involuntary movements, and Kayser-Fleisher rings in her eyes on slit lamp examination.

Serum ceruloplasmin was 8mg/dl (normal range: 20-40mg/dl), serum-free copper was 125mg/dl (normal range 10-15mcg/dl), alkaline phosphatase 113 IU.

Her MRI brain suggested a symmetrical altered signal Intensity lesion in bilateral thalami and lentiform nucleus, midbrain and pons, favouring a diagnosis of Wilson's disease. She was started on the treatment of steroids, zinc, syndopa and D-penicillamine, which she was not compliant with.

Presently, she is consciously oriented to time, place and person. She has slurring of speech, flexion deformity in bilateral hip and knees, weakness in all 4 limbs and kyphosis in the thoracic spine. Neuropsychiatric changes manifested as grimaces and facial grinning.

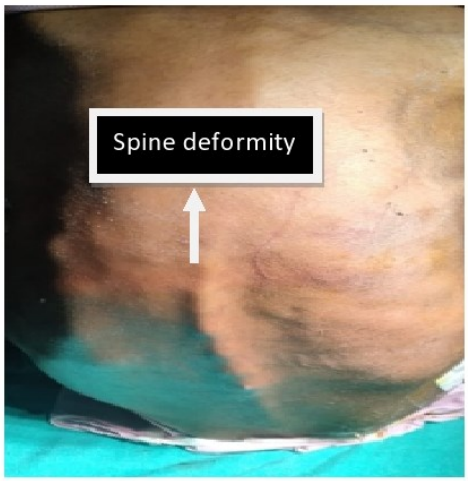

Fig. 1A: Spine defomity

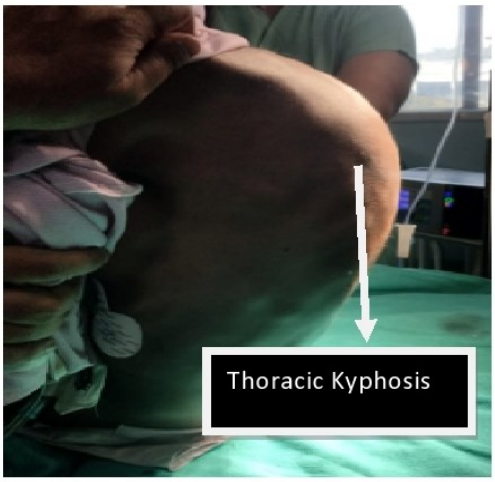

Fig. 1B: Thoracic kyphosis

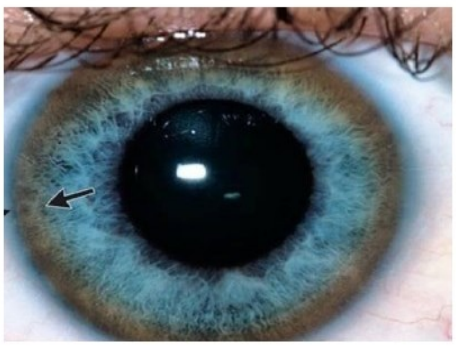

Fig. 2: Kayser Fleisher rings

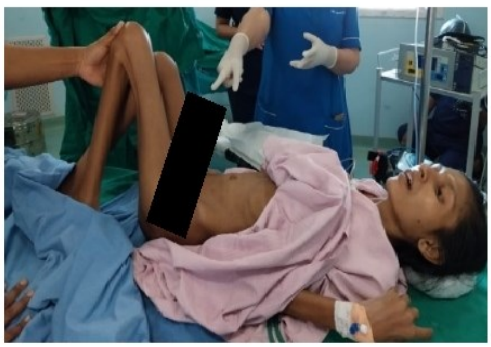

Fig. 3: Fixed flexion deformity in lower limb

Her haemoglobin was 9.6 gm/dl, alkaline phosphatase level was 643 IU/L (normal range: 30-120 IU), INR- 1.2, Vitamin D3 level 3ng/ml, for which correction was started. The rest of blood investigations were within normal limits. Ultrasound abdomen suggested a heterogenous liver. 2D echocardiography was normal.

She was re-started on D-penicillamine and zinc. Rapport was made with the patient pre-operatively and parents were counselled about the peri-operative risk of surgery and anaesthesia.

On the day of surgery, her pulse rate was 125/min regular in rhythm, her blood pressure was 110/60 mm of Hg, and her saturation was 100% in the room air. Our anaesthetic technique was a combined spinal epidural. As patient positioning was challenging due to the kyphosis, pain at the fracture site and fixed flexion deformity, we gave 12cc of 0.125% Bupivacaine in ultrasound-guided infra-inguinal fascia iliaca block. Combined spinal-epidural was administered with 10 mg of 0.5% (H) Bupivacaine injected into the

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subarachnoid space at L3-L4 interspace and the epidural was secured at L2-L3 level. Adequate blockade was achieved till T10. The patient's vitals were monitored. As the patient was anxious, we started injection dexmedetomidine infusion at 0.2mcg/kg for 10 mins (6mcg of inj. Dexmedetomidene), which led to an apnoeic episode and the patient desaturated to 85%, heart rate dropped from 120 bpm to 60 bpm. Dexmedetomidine infusion was immediately stopped, and Atropine 0.6mg iv and IPPV were given with 100% oxygen for 5 mins. Heart rate increased to 130/min. Once spontaneous efforts resumed, Hudson Mask was used with side stream capnography to monitor respiration.

Intra-operative blood loss was 200ml, Input was 1100ml, and urine output was 300ml. Epidural top-up with 6 mL of 0.0625% of Bupivacaine with 30 µg of Buprenorphine was given. Post-operatively, the patient was observed in MICU where one pint of packed red cells was transfused. Analgesia was maintained with injection of tramadol 50 mg, 12th hourly and 8th hourly epidural top-ups. On day 3, a repeat fracture of the distal femur on the same side was noted by orthopedicians on an X-ray. The epidural catheter was removed on day 5 after a discussion with orthopedicians. Tab D- penicillamine was restarted.

On postoperative day 7, the patient was posted for plating of the distal femur. Repeat investigations were normal. VitD3 was corrected to 63 IU. Combined spinal-epidural was given, facilitated by ultrasound-guided preoperative femoral nerve block. Considering the earlier episode of apnoea, no sedatives were given this time. The entire perioperative period concluded with the patient's gratifying smile and a sweet thank you.

DISCUSSION

Wilson's disease is a rare autosomal recessive disorder characterized by hepatic, ophthalmic, and neuro-psychiatric symptoms from excessive copper accumulation. Genetic testing of 1st& 2nd degree relatives is essential for rapid diagnosis of prevalent mutations. The copper level needs to be optimised by starting pharmacotherapy.

Our patient had contractures, slurring of speech and behavioural abnormalities associated with parkinsonism caused by copper deposits in basal ganglia and hippocampal regions. Bulbar symptoms included speech problems, dysphagia and salivary drooling. Psychiatric signs included depression and behavioural abnormalities. Kayser-Fleisher rings [2], a classical ophthalmological finding of Wilson's disease was seen.

Acute liver failure, cirrhosis, and modest histological alterations are examples of hepatic impairment. Patients can present with diastolic dysfunction, cardiomyopathy, conduction abnormalities and ventricular arrhythmias [3]. The hemodynamic reserve may deteriorate with autonomic dysfunction [4], which bedside tests should rule out.

There may be respiratory failure [5] due to weak muscles. Hypnotic and sedative drugs can exacerbate neurological, psychiatric problems and cause respiratory depression. Even mild sedation may result in apnoea and respiratory depression, which was reported intra-operatively in our patient with just 6 mcg of dexmedetomidine [6,7] over 10 minutes.

There are only a few case reports of anaesthetic management of Wilson's disease. Cases have been reported under general anaesthesia [8,9], neuraxial anaesthesia [10,11] and regional block [12], techniques. Regional anaesthesia decreases the use of systemic analgesics like opioids, which need hepatic metabolism and cause neurological impairment. General anaesthesia is disadvantageous as it may worsen the already impaired liver function. Sensitivity to muscle relaxants is increased secondary to D-penicillamine and elevated copper levels per se. After documenting the pre-existing neuromuscular weakness assessed by neuro-physician, we employed regional anaesthesia in our patient. These patients are severely osteoporotic [13]and may be associated with metabolic bone disease [14], so careful shifting is required. Post-operative sepsis [15,16] should be watched for along with worsening neuropsychiatric symptoms [17,18].

CONCLUSIONS

Wilson's disease is a challenge to anaesthesiologists. Appropriate management requires a multidisciplinary approach utilizing the expertise of an anaesthesiologist, physician, orthopedician, and neurologist and detailed knowledge of the organ systems involved. The complications differ with each case and so does the management.

As there is a lack of literature on anaesthetic management of Wilson's disease due to the rarity of this condition, we hope to increase the database by

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publishing our case report. An international registry of this disease should be maintained so that peri-operative clinicians are well informed about the peri-operative challenges and patient care can be par excellence. Informed consent was obtained from the patient and her relatives to publish this case report.

CONTRIBUTION OF AUTHORS

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Research design- Not applicable

Supervision- Dr. Heena Pahuja, Dr. Prerna Agrawal, Dr. Anjali Bhure

Materials- Not applicable

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Final approval – Dr Heena Pahuja, Dr Prerna Agrawal, Dr Anjali Bhure

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