CASE REPORT

Patient with ataxia telangiectasia undergoing elective staging laparoscopy: A case report and literature review

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Abstract

Ataxia telangiectasia is a rare autosomal recessive condition which develops due to a mutation in the ataxia telangiectasia mutated gene (ATM gene). As a result of this mutation, the ability of the DNA to undergo repair is undermined. The resulting cellular demise is responsible for the diverse presentation of the clinical condition. Neurological symptoms such as cerebellar ataxia, abnormal eye movements and malignancies occur commonly, while immunodeficiency predisposes these patients to recurrent infections. Perioperative management of patients with this rare condition can be associated with increased morbidity. Therefore, it is recommended that patients with ataxia telangiectasia should be managed in a multidisciplinary centre, under the supervision of senior clinicians who have the insight into the clinical needs of such patients. We report herein, the perioperative management of a patient with ataxia telangiectasia undergoing laparoscopic procedure. This case report will allow the readers to increase their knowledge and understanding when it comes to the management of these patients.

Keywords: Ataxia Telangiectasia Syndrome, Ataxia Telangiectasia, Louis-Bar Syndrome

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Introduction

Ataxia telangiectasia (A T), also called Louis-Bar Syndrome, is a rare autosomal recessive condition.[1] It is caused by a mutation in the ATM (Ataxia Telangiectasia Mutated) gene, leading to impairment of the mechanism responsible for DNA repair. The prevalence of this condition ranges from 1-2.5 per 100,000 live births with average life expectancy of 25 years,[2] affecting both male and female genders equally.[3]

The clinical picture of this rare condition includes a range of neurological conditions, skin involvement, increased susceptibility to infections due to immunodeficiency and a predisposition towards developing various malignancies, particularly of lymphoid origin.[2,4]

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Due to unfamiliarity with the diagnostic criteria or the signs and symptoms of this rare condition, clinicians often find it difficult to correctly diagnose ataxia telangiectasia. It may often be confused with cerebral palsy, congenital ocular motor apraxia and Friedreich's ataxia. One method of confirming the diagnosis of ataxia telangiectasia is to identify the mutations in the ATM gene. It can also be diagnosed by determining the absence or deficiency of ATM protein and/or ATM kinase activity in the cell lines cultured from lymphocytes or skin biopsies.[4]

Case Report

A 22-year-old male, weighing 45 kg, American Society of Anaesthesiologist Physical Status (ASA-PS) grade 3, with primary malignancy of gastroesophageal junction, was planned for elective staging laparoscopy at Shaukat Khanum Memorial Cancer Hospital and Research Centre Lahore, on February 2019.

The patient was a known case of ataxia telangiectasia. He had been wheelchair-bound since birth; he was able to sit, however, could not bear weight on his legs. Six months earlier, the patient had developed left sided chest pain. It was appropriately investigated by the cardiology team and no cardiac issues were detected. The patient was referred to gastroenterology for upper GI endoscopy, which revealed the source of the chest pain to be a gastroesophageal tumour. Since, two months, the patient's food intake had decreased as he had not been able to take solid food and had been relying on semisolid and liquid diet. This led to a marked weight loss. The patient had also experienced speech difficulties for the last two months. He had no other co-morbid conditions.

Family history revealed that the patient had three other siblings who also had ataxia telangiectasia and had passed away.

On examination, excessive skin pigmentation was present on the sun-exposed regions of the body. He appeared to be malnourished and his chest revealed pectus carinatum. The auscultatory examination of his chest was otherwise unremarkable.

Neurological examination revealed decreased power in both the lower limbs,[5,6] while power in the upper limbs was normal. There was no sensory deficit. Deep tendon reflexes

were also normal. Examination of the spine revealed scoliosis.

Examination of the airway showed the mouth opening to be two finger breadths, thyromental distance was less than 6cm, neck extension was adequate, mallampati classification was grade 2, prominent incisors and multiple other teeth were missing.

The patient had no known allergies. It was not possible to assess the patient's functional status. His current medications included Omeprazole and Paracetamol tablets.

After standard monitoring was instituted and intravenous access had been established, the patient was pre-oxygenated. Intravenous induction was done with Morphine 3mg, Propofol 40mg followed by Atracurium 25 mg.

C-MAC video laryngoscope was used to intubate the patient with a size 7.0 endotracheal tube. Anaesthesia was maintained with 50% oxygen, 50% air and Sevoflurane. Antibiotic Cefazolin 1gm was given intraoperatively. Injection Dexamethasone 4mg and Ondansetron 4mg and Paracetamol 700mg were given intravenously. At the end of the procedure, the surgical team infiltrated the port insertion sites with 10ml of 0.5% bupivacaine. Neuromuscular blockade was reversed with Neostigmine and Glycopyrrolate combination, and the patient was successfully extubated.

Throughout the procedure the patient's temperature was monitored and active warming methods were used. This procedure was continued in the post anaesthesia care unit (PACU) with the aim to keep the patient normothermic.

The patient remained haemodynamically stable and pain free in PACU, and no complications were reported. He was transferred to the ward after 50 minutes stay in the PACU and remained admitted in the ward overnight. He was discharged the next day.

Discussion

Ataxia telangiectasia develops due to mutations of the ATM gene and the resulting lack of repair of the DNA leads to cell death of the tissues, which may lead to diverse and complex presentation of this condition, especially affecting the brain, causing neurological symptoms such as cerebellar ataxia and abnormal eye movements during early life. Moreover, development of malignant proliferation is also a common occurance.[1,3]

Ataxia is the first noticeable sign; it can be noticed in toddlers as unstable gait or instability of the trunk. Patients may become wheelchair bound by 10 years of age. Further progression of the disease can lead to dysarthria, oculomotor apraxia, nystagmus, saccadic intrusions and telangiectasias.[1]

Basal ganglia involvement presents as tremor, chorea, dystonia, myoclonus and parkinsonism. Axonal neuropathy may be responsible for the various orthopaedic abnormalities commonly seen in these patients.[1]

Telangiectasia can also commonly occur in the conjunctiva and/or in the skin exposed to the sun.[5] They can also develop in the bladder tissues.[5] Telangiectasias do not evolve nor do they bleed. These patients can also develop cutaneous greying and premature aging.[1]

These patients also suffer from immunodeficiency which predisposes them to develop recurrent sinopulmonary infections. Recurrent chest infections along with restrictive pulmonary defect can lead to development of bronchiectasis and interstitial lung disease.[8]

Twenty-five to 30% of the patients are prone to develop tumours. Leukaemia and lymphomas arise early in life and solid tumours such as breast, melanomas, ovarian, liver and gastric tumours can develop later in life.[1]

According to some evidence, anaesthesia for patients with ataxia telangiectasia can be associated with increased morbidity. Due to the prevalence of interstitial lung disease, increased risk of aspiration, infection and decreased lung compliance, anaesthesia in these patients is associated with the same risks as any other medically complex patient.[7]

Management of patients with ataxia telangiectasia should ideally be done in multidisciplinary care centre/hospital. This would ensure that the patient receives disease specific perioperative optimisation and postoperative follow up. Pre-operative testing (pulmonary function tests, X-rays) should be performed if clinically indicated.[2]

Nutritional status is an important indicator of the status of the disease in these patients. However, declining motor coordination leads to feeding and communication difficulties. Therefore, every effort should be made to maximise nutritional support before and after surgery.[2]

Perioperative risk for patients with ataxia telangiectasia is reduced with an increase in the experience of the physicians involved in their management. The availability of experienced consultants, and intensive care facilities all contribute to better patient outcome.[2]

Conclusion

Morbidity in patients with ataxia telangiectasia can be high if they are inappropriately managed in the perioperative period. Therefore, it would be reasonable to say that before embarking on surgery, these patients should be thoroughly evaluated by an experienced multidisciplinary team. Appropriate investigations should be carried out. Close monitoring of the patient should be continued both intraoperatively and in the post-operative period. If mechanical ventilation becomes necessary in the immediate post-operative period, all efforts should be made to wean the patient from mechanical ventilation as early as possible.

Consent: Before submission for publication, this case report was reviewed by the Institutional Review Board of Shaukat Khanum Memorial Cancer Hospital and Research Centre, Lahore. IRB approval was taken from the institution and a consent was obtained from the patient at the time of admission regarding the usage of data for research and educational purposes.

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References

1. Riboldi GM, Samanta D, Frucht S. Ataxia Telangiectasia (Louis-Bar Syndrome). Treasure Island FL: StatPearls Publishing, 2018.
2. Lockman JL, Iskander AJ, Bembea M, Crawford TO, Lederman HM, Morrow SMG, et al. Anaesthetic and preoperative risk in the patient with Ataxia-Telangiectasia. Paediatr Anaesth. 2012; 22:256-62.
3. Bhatt JM, Bush A, Gerven M, Nissenkorn A, Renke M, Yarlett L, et al. Ataxia telangiectasia: why should the ERS care? Eur Respir J. 2015; 46:1557-60.
4. Rothblum-Oviatt C, Wright J, Lefton-Greif MA, McGrath-Morrow SA, Crawford TO, Lederman HM. Ataxia telangiectasia: a review. Orphanet J Rare Dis. 2016; 11:159.
5. Greenberger S, Berkun Y, Ben-Zeev B, Levi YB, Barziliai A, Nissenkorn A. Dermatologic manifestations of ataxia-telangiectasia syndrome. J Am Acad Dermatol. 2013; 68:932-6.
6. Lin DD, Barker PB, Lederman HM, Crawford TO. Cerebral abnormalities in adults with ataxia-telangiectasia. AJNR Am J Neuroradiol. 2014; 35:119-23.
7. Van Os NJ, Haaxma CA, Flier MV, Merkus PJ, Deuren MV, Groot IJ, et al. A-T Study Group. Ataxia-telangiectasia: recommendations for multidisciplinary treatment. Dev Med Child Neurol. 2017; 59:680-9.
8. McGrath-Morrow SA, Lederman HM, Aherrera AD, Lefton-Greif MA, Crawford TO, Ryan T, et al. Pulmonary function in children and young adults with ataxia telangiectasia. Pediatr Pulmonol. 2014; 49:84-990.