

Tuberous sclerosis- An anaesthetic challenge

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Abstract

Tuberous sclerosis is a relatively rare genetic syndrome. Awareness of the signs and symptoms and the organs involved is critical to provide safe & effective anesthetic care. We hereby describe a case of a 10 year old boy scheduled for routine Magnetic resonance imaging tumor screening of abdomen, pelvis and chest. This child was severely mentally retarded, autistic, physically challenged & gave history of seizures for which he was on anticonvulsant therapy.

Keywords: Anaesthesia, Tuberous sclerosis, Seizures, Mental retardation

Introduction

Tuberous sclerosis also known as Bournville's disease is a rare autosomal dominant genetic disorder. It is characterized by the classical triad of mental retardation, seizures and facial angiofibromas. Tuberous sclerosis occurs in approximately 1:6000 to 1:10000 live births and is usually diagnosed during the first decade of life^[1,2]. There is large variation in the severity of the disease^[2,3]. People with mild disease have normal life expectancy while children with severe form of this disease might die in early childhood^[4].

Systemic manifestations of tuberous sclerosis may include seizures, mental retardation, facial angiofibromas, ash leaf spots, giant cell astrocytomas (GCA), angiomyolipomas of kidney, rhabdomyomas of the heart, adenoma of the liver, lymphangiomyomatosis in the lungs. Skeletal muscles, peripheral nerves & the spinal cord are the only tissues unaffected by tuberous sclerosis tumor growth^[3,4,5].

Case Report

A 10 year old boy weighing 25 kgs presented to the radiology department of our institute for routine Magnetic Resonance Imaging (MRI) tumor screening of abdomen, pelvis and chest. The child had been diagnosed with tuberous sclerosis at the age of

4 years. The child appeared severely mentally retarded, autistic & was physically handicapped. The child was unable to respond to physical and verbal communication. The child's parents gave history of seizure disorder and was receiving lamotrigine medication for the same.

On pre-anaesthetic evaluation, the airway was difficult to assess as the child was unco-operative. There appeared no limitation of child's neck movements. Cardio-vascular and respiratory system examinations were normal. Pre-operative routine blood investigations were within normal limits. Electro-cardiogram (ECG) was normal.

The child was brought to MRI suite. Standard ASA monitors compatible with MRI were applied. Intravenous access was achieved with 22G intravenous cannula and dextrose normal saline was started. We planned for general anaesthesia with controlled ventilation with appropriate size endotracheal tube, avoiding hypoventilation and trauma to the airway. The child was pre-medicated with inj. Glycopyrrolate 4µg/kg, ondansetron 0.15mg/kg, midazolam 0.03mg/kg. The patient was pre-oxygenated with 100% oxygen for 5min. Induction was done with inj. Propofol 50mg and Atracurium 10mg to facilitate endotracheal intubation. Anaesthesia was maintained with

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isoflurane and nitrous oxide in oxygen. The complete imaging study lasted for 2hrs 30min. After completion, the neuromuscular blockade was reversed with inj. Neostigmine 1.25mg and inj. Glycopyrulate 0.2mg. The patient was carefully extubated after thorough oropharyngeal suction and return of appropriate spontaneous respiration. The patient was shifted to post-anaesthesia care unit and kept under observation with supplementary oxygen for 30min.

Discussion

Limited information is available in the literature regarding anaesthetic management of patients with tuberous sclerosis. Pre-anaesthetic assessment of the patient should focus both on the expected abnormalities secondary to the disease process as well as presence of any co-morbid conditions. Neurological status of the patient may affect his ability to co-operate.

Airway assessment should be made to rule out the presence of oral lesions which can cause difficulty in securing airway. Patients with tuberous sclerosis may manifest with fibroma, papilloma and nodular tumors on the tongue and palate (11%). Mass ventilation may be difficult especially when these tumors are located in posterior pharynx following induction of anaesthesia resulting in unmasking of airway obstruction^[6]. Cardiovascular system assessment should be made carefully as 50% of children with tuberous sclerosis will develop rhabdomyoma of the heart which may cause congestive cardiac failure, conduction abnormalities, refractory arrhythmias and severe haemodynamic compromise^[7]. It is necessary to obtain an ECG and possibly an echocardiogram if there is suspicion of cardiac involvement.

Patients with tuberous sclerosis often are mentally retarded which may range from mild to severe retardation. 3 brain lesions most common are subependymal nodules, cortical tubers & giant cell astrocytoma(GCA)^[3]. These 3 neoplasms are believed to cause mental retardation, autism, hyperactivity disorder and seizures^[8,9].

Anaesthesia care may be complicated by behavioural disturbances in unco-operative patients who lack the ability to communicate. Patients often are on medication for seizure control which increases the risk for drug-induced interactions or adverse

reactions. General anaesthesia with controlled ventilation is safe anaesthesia to keep the patient euvoletic and normocarbic. High or low carbon dioxide levels may increase the potential for a seizure^[10].

Choice of neuromuscular blocker depends on the hepatic and renal function of the patient. If patient has decreased liver function, rocuronium may be of better choice than vecuronium as it is primarily metabolized by the liver^[11,12]. Renal function may be compromised secondary to renal hamartomas or polycystic disease. In such cases, atracurium is the choice of neuromuscular blocker as it is not renally excreted^[10,11,12]. In our case, we used atracurium as neuromuscular blocker as it is safest with no hepatic metabolism and renal excretion.

Respiratory system is rarely involved in tuberous sclerosis. Less than 1% patients present with direct lung involvement. Two pulmonary conditions which may be associated with tuberous sclerosis are lymphangioleiomyomatosis and Multifocal Micronodular Pneumocyte Hyperplasia (MMPH)^[13,14,15]. Lymphangioleiomyomatosis causes interstitial smooth muscle proliferation that leads to interstitial thickening, alveolar damage & finally chronic fibrosis of the lung. In MMPH, multiple nodules are formed from alveolar septa and type II alveolar cells proliferate^[16,17]. Such patients may present with dyspnoea, haemoptysis, pulmonary hypertension and finally cor-pulmonale^[1,15,16,17]. Patients with severe form of disease may develop recurrent pulmonary infection and so anaesthesiologist should rule out pneumonia or presence of any other infection. Chest X-ray should be obtained to rule out any pulmonary involvement^[10].

Pre-anaesthetic evaluation of our patient did not reveal any evidence of cardiac or pulmonary disease but neurological involvement was severe as child was severely mentally retarded and was unable to co-operate or comprehend. This may potentially affect the choice of technique for establishing general anaesthesia. Thus, as an anaesthesiologist peri-operative management of patients with tuberous sclerosis is often complicated by presence of cardiovascular, neurological and renal tumors. Pre-anaesthetic evaluation of the patient should be carefully done to rule out involvement of each system their extent and severity of the disease.

References

1. Weiner DM, Ewalt DH, Roach ES, Hensle TW. The tuberous sclerosis complex: a comprehensive review. *J Am Coll Surg* 1998;187:548-61.
2. Dinno ND, Griffith's 5-minute Clinical Consult. 10th ed. Philadelphia, Pa-Lippincott Williams & Wilkins; 2002:1124-25.
3. Lendvay TS, Marshall FE. The tuberous sclerosis complex & its highly variable manifestations. *J Urol* 2003;169:1635-42.
4. Byard RW, Blumbergs PC, James RA. Mechanisms of unexpected death in tuberous sclerosis. *J Forensic Sci* 2003;48:172-6.
5. O'Callaghan FJ. Tuberous sclerosis: epidemiological research is needed to complement new findings in genetics. *BMJ* 1999;318:1019-1020.
6. Gomez MR. History of the tuberous sclerosis complex. *Brain Dev* 1995;17:55-57.
7. Behrman, in Nelson Textbook of Pediatrics, 17th ed., 2004, Ch. 589 Neurocutaneous Syndromes, Philadelphia PA, WB Saunders, pp. 1705- 9.
8. Cuccia V, Zuccaro G, Sosa F, Monges J, Lubienieky F, Taratuto AL. Subependymal giant cell astrocytoma in children with tuberous sclerosis. *Childs Nerv Syst* 2003;19:232-43.
9. Lee JJ, Imrie M, Taylor V. Anaesthesia & tuberous sclerosis. *Br J Anaesth* 1994;73:421-25.
10. Diaz JH. Perioperative management of children with congenital phakomatoses. *Pediatr Anesth* 2000;10:121-28.
11. Cote CJ, Todres ID, Ryan JF, Goudsouzian NG. *A Practice of Anaesthesia for Infants & Children*. 3rd ed. Philadelphia, Pa: WB Saunders; 2001;45:184-185.
12. Morgan GE, Mikhail MS, Murray MJ. *Clinical Anesthesiology*. 3rd ed. New York, NY: McGraw-Hill; 2002;857-862.
13. Johnson SR, Clelland CA, Ronan J, Tattersfield AE, Knox AJ. The TSC-2 product tuberini sex pressed i n lymphangioliomyomatosis & angioliopoma. *Histopathology* 2002;40:458-63.
14. Matsumoto S, Nishioka T, Akiyama T. Renal angiomyolipoma associated with micronodular pneumocyte hyperplasia of the lung with tuberous sclerosis. *Int J Urol* 2001;8:242-44.
15. Cancellieri A, Poletti V, Corrin B. Respiratory failure due to micronodular type II pneumocyte hyperplasia. *Histopathology* 2002;41:263-65.
16. Sullivan EJ. Lymphangioliomyomatosis. *Chest* 1998;114:1689-1703.
17. Lantuejoul S, Ferretti G, Negoescu A, Parent B, Brambilla E. Multifocal alveolar hyperplasia associated with lymphangioliomyomatosis in tuberous sclerosis. *Histopathology* 1997;30:570-75.

Source of Support : **Nil**

Conflict of Interest : **None Declared**