

Tuberous Sclerosis: A Case Report and Review

E Bryson, S Neustein

Citation

E Bryson, S Neustein. *Tuberous Sclerosis: A Case Report and Review*. The Internet Journal of Anesthesiology. 2006 Volume 13 Number 1.

Abstract

Tuberous sclerosis, also known as Bourneville's disease, is an autosomal dominant genetic disorder classically characterized by the triad of mental retardation, seizures, and facial angiofibromas. The small benign tumors in the brains of affected individuals are the cause of the neurologic manifestations of the disorder, and often develop in the visceral organs as well. Routine screening of patients with tuberous sclerosis to determine if involvement of secondary organs has occurred is accomplished via magnetic resonance imaging, which almost always requires general anesthesia. We report the management of a patient with tuberous sclerosis complicated by severe mental retardation and autism undergoing magnetic resonance imaging (MRI) for tumor surveillance.

CASE REPORT

A 14 year old, 40 kg Hispanic male with tuberous sclerosis presented for routine MRI tumor screening of the chest, abdomen and pelvis. He had been a ward of the state since being diagnosed with tuberous sclerosis at age 2, and was severely mentally retarded, autistic, and physically handicapped, being fully dependent on others for care. He was unable to communicate with his caregivers directly and did not respond to attempts at verbal or physical communication. At baseline, his routine consisted of auto stimulation of the hands and face with occasional "whooping" sounds. Both his mother and the caregiver who accompanied him to the hospital described his demeanor as generally calm but unresponsive. His seizure disorder was controlled with divalproex, and he was routinely administered topiramate and levocarnitine as well. He had no history of allergy to foods or medications and no prior anesthetic history.

On physical exam the patient continued his auto stimulatory movements and did not respond to physical contact. His airway was difficult to assess due to his inability to cooperate, but his caregiver reported no loose or missing teeth. His thyromental distance was 6.5 cm and there appeared to be no limitations on his neck range of motion. His pulmonary and cardiac exams were both unremarkable. Though he did not exhibit contractures, his arms were tightly flexed and returned to this position if straightened. The patient allowed manipulation of his arms so long as the limbs were moved slowly. Pre-operative chemistries and

ECG were unremarkable.

The patient was brought to the MRI suite in his wheelchair, unaccompanied by his caregiver. He was moved from the chair to the MRI table and brought into the room without difficulty. Standard ASA monitors compatible with MRI were applied and several attempts were made to locate an appropriate site for intravenous (IV) cannulation. The patient tolerated the attempts at IV placement without responding. Since these veins were extremely small and were not successfully cannulated, an inhalational induction of anesthesia was performed. Sevoflurane (8%), nitrous oxide (7L) and oxygen (3L) were administered via facemask and tolerated well by the patient. Once induction of anesthesia had been achieved, a 20 gauge intravenous catheter was placed in the patients left forearm without difficulty. Vecuronium 5 mg and propofol 50 mg was administered to facilitate tracheal intubation and ventilation and anesthesia was maintained with isoflurane and nitrous oxide in oxygen. The imaging study lasted two hours and forty minutes, after which the neuromuscular blockade was reversed with neostigmine (2mg) administered with glycopyrrolate (0.4mg). Once spontaneous respirations had resumed and after the oropharynx was suctioned, the endotracheal tube was removed and the patient was brought to the post anesthesia care unit without incident.

DISCUSSION

There is limited information available in the literature regarding the anesthetic management of patients with

tuberous sclerosis. Case reports are limited to specific surgeries, with the majority in non-English journals. ¹ One report of general anesthesia for a patient undergoing two-stage scoliosis surgery exists in the British literature ² and a more recent report of a 10-year-old girl with tuberous sclerosis who received a general anesthetic for laser treatment of facial angiofibromas, ³ but a comprehensive literature search revealed no reports of anesthesia for magnetic resonance imaging studies.

Patients with tuberous sclerosis commonly develop angiomyolipomas. Common sites for these tumors include the kidneys, lungs, and the heart. ⁴ Up to 50% of children with tuberous sclerosis will develop rhabdomyomas of the heart, which may cause congestive heart failure, conduction abnormalities, refractory arrhythmias, and severe hemodynamic compromise. ⁵ When the kidneys are involved, the typical presentation is hematuria, pain, and, in some cases, renal failure secondary to hamartomas or polycystic disease. Lung involvement by angiomyolipomas that produce generalized cystic or fibrous pulmonary changes can lead to spontaneous pneumothorax, which may lead to tension pneumothorax under positive pressure ventilation.

The most common neurologic manifestations of tuberous sclerosis include mental retardation, which can be severe and incapacitating, and seizures. ⁶ About 60% of affected individuals exhibit some degree of delayed mental development ranging from mild to severe mental retardation, and 25% to 60% will also meet the diagnostic criteria for autism. ⁷ Almost all patients with tuberous sclerosis present with pervasive developmental disorder. ⁸ Anesthesia care may be complicated by behavioral disturbances in the uncooperative patient who lacks the ability to communicate and cannot be consoled. Placement of intravenous access for induction of anesthesia may not be possible, and an inhalation induction may be required.

The preanesthetic assessment of the patient with tuberous sclerosis should focus both on the expected abnormalities secondary to the disease process as well as the presence of any co-morbid conditions. The neurologic status of the patient may affect the ability of the patient to cooperate, and the type of anesthesia which may be possible. It is the rare individual with tuberous sclerosis who does not require general anesthesia for surveillance MRI. Seizures may not be well controlled, and the patient may be taking a number of different anti-seizure medications which increases the risk

for drug-drug interactions or adverse reactions.

The airway exam should note the presence of oral lesions as these can make airway management difficult. Nodular tumors, fibromas, and papillomas have been found on the tongue and palate in 11% of patients with tuberous sclerosis. ⁹ When located in the posterior pharynx, the induction of anesthesia may unmask airway obstruction, resulting in a patient who is difficult, or even impossible to ventilate.

The cardiac status of the patient may be difficult to evaluate by history, especially if the patient does not communicate verbally. It may be necessary to obtain an ECG, and possibly an echocardiogram if there is suspicion of cardiac involvement.

The respiratory system is rarely impaired, with less than 1% of patients presenting with direct lung involvement due to tuberous sclerosis. ¹⁰ Frequently patients with the severe form of the disease develop recurrent pulmonary infections and care should be taken to rule out the presence of any pneumonia or other infection in the patient who is undergoing an elective procedure. Any history of pneumothorax raises concerns for the development of a tension pneumothorax under positive pressure ventilation, and a chest x-ray should be obtained if there is any suspicion. When they are present, pulmonary lesions have a poor prognosis and are often result in the death of the patient. ¹¹

Evidence of involvement of the renal system can be elicited through history and the evaluation of a basic metabolic panel or urinalysis. Symptoms are generally mild and the patient will often complain of a vague, poorly localized abdominal, lower back or flank pain. Occasionally acute renal failure is the presenting symptom of tuberous sclerosis in young children. ¹²

A review of our patient's medical history did not reveal any evidence of systemic cardiac or pulmonary disease which would have altered our management. His ECG was without evidence of ischemic changes or prior myocardial infarction, and preoperative chemistries were normal. The extent of his neurological compromise, however, was severe, presenting us with a patient with no ability to understand or cooperate. This could potentially affect the choice of techniques for establishing general anesthesia, especially if the patient had become combative. We also had to rely on our clinical judgment that he was awake and strong, without any ability of the patient to follow commands, to determine when

extubation could be accomplished safely.

The prognosis for the patient with tuberous sclerosis depends on the severity of symptoms. Although individuals with more severe symptoms may have serious disabilities, the life expectancy is not significantly decreased given appropriate medical care.¹³ Anesthesiologists can expect that patients with tuberous sclerosis, though rare, may present at any age and with a wide range of morbid and co-morbid conditions.

CORRESPONDENCE TO

Steven M. Neustein, M.D. Mount Sinai Medical Center 1
Gustave L. Levy Place Box 1010, Dept. of Anesthesiology
New York, NY 10029-6574 Telephone (212) 241-7467 Fax:
212-426-2009, and Email: steve.neustein@msnyuhealth.org

References

1. Mimura S, A case of a pediatric patient with tuberous sclerosis (Bourneville-Pringle disease) and frequent epileptic seizure for intensive dental treatment under general anesthesia, Masui (Japanese Journal of Anesthesiology) (2006) 55(4): pp. 471-474
2. Lee JJ, Imrie M, Taylor V, Anaesthesia and tuberous sclerosis, British Journal of Anaesthesia, (1994) 73: pp.421-425
3. Septer S, Thompson ES, Willemsen-Dunlap A, Anesthesia concerns for children with tuberous sclerosis, AANA journal. (2006) 74(3): pp. 219-225
4. Behrman, in Nelson Textbook of Pediatrics, 17th ed., 2004, Ch. 589 Neurocutaneous Syndromes, Philadelphia PA, WB Saunders, pp. 1705-1709.
5. Cope J.T., Lindsey J.H., Kron I.L., Cardiac tumors. C.C.L. Pediatric cardiac surgery, 3rd ed, 2003, St. Louis: Mosby, pp 689-701.
6. Curatolo P., Verdecchia M., Bombardieri R., Tuberous Sclerosis complex: A review of neurological aspects. Eur J Paediatr Neurol (2002) 6: pp 15-23.
7. Ridler K, et al, Neuroanatomical Correlates of Memory Deficits in Tuberous Sclerosis Complex, Cerebral Cortex (2007) 17(2):261-271
8. Harrison JE, Bolton, PF, Annotation: Tuberous sclerosis, Journal of Child Psychology and Psychiatry (1997) 38: pp 603-614.
9. Gomez MR, ed. Tuberous Sclerosis, 2nd Ed., New York, Raven Press, 1988, pp. 139-148
10. Lie JT, Cardiac, pulmonary, and vascular involvements in tuberous sclerosis, Annals of the New York Academy of Sciences (1991) 68: pp 58-70.
11. Shepherd CW, Gomez MR, Lie JT, Crowson CS, Causes of death in patients with tuberous sclerosis, Mayo Clinic Proceedings, (1991) 66: pp. 792-796
12. Bernstein J, Robbins TO, Renal involvement in tuberous sclerosis, Annals of the New York Academy of Sciences, (1991); 615: pp. 36-49
13. Shepherd C, Gomez M, Lie J, Crowson C, Causes of death in patients with tuberous sclerosis, Mayo Clin Proc (1991) 66 (8): 792-6.

Author Information

Ethan Bryson, M.D.

Instructor, Department of Anesthesiology, The Mount Sinai Hospital

Steven M. Neustein, M.D.

Associate Professor of Anesthesiology, Department of Anesthesiology, The Mount Sinai Hospital