Management of a patient with osteogenesis imperfecta: A case study

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The pathophysiological manifestations of osteogenesis imperfecta, a rare disease of the connective tissue, require special anesthetic considerations. The case study presented in this article illustrates the anesthetic management of a patient with this disorder.

Osteogenesis imperfecta was formerly thought to be a disease seen only by pediatricians and orthopedic surgeons, since most of the afflicted patients died before achieving sexual maturity. With improved health care, however, patients with osteogenesis imperfecta are now reaching reproductive age and are being encountered by both the obstetrician and the obstetrical anesthetist.

Osteogenesis imperfecta is a rare and inherited disease of the connective tissue with the skeletal tissue being the most affected. Malebranche first described osteogenesis imperfecta in 1678. In 1906, Losser classified two variants: congenita, acquired in utero, and tarda, the more common variant seen in childhood.

Transmitted as a Mendelian dominant, the occurrence of the osteogenesis imperfecta gene is approximately 50%, with females having a slightly higher incidence than males. The incidence in the general population is 1:20,000 to 1:60,000, with an estimated 4,000 patients living in the United States.

Physical characteristics include blue sclera in over 90% of the cases, deafness secondary to otosclerosis in 25-50% of the cases, and a greatly increased susceptibility to skeletal fractures. The frequency of fractures tends to decrease after puberty, only to reappear, especially in menopausal females. Other characteristic symptoms include a triangularly shaped face, a short neck, spinal osteoporosis, thin skin and joint hypermobility. Deformity of the thorax in some patients can be the cause of respiratory insufficiency. McKusick has reported emphysema and arteriosclerosis at an early age. In general, the teeth are very fragile and more susceptible to caries.

There is no known cure for osteogenesis imperfecta, nor has any pharmacologic regimen been shown to be beneficial. Deformed limbs can be straightened surgically with medullary nails in the hope that they will remain functional until adolescence brings remission of the disease process. Other anomalies which may accompany osteogenesis imperfecta are hydrocephalus, congenital heart defects, hemophilia and cleft palate.

Of great interest to the anesthetist in the clinical setting are the following laboratory findings: an increased basal metabolic rate and a decreased platelet aggregation. Gebala has reported an increase in serum acid phosphatase. Although frequent epistaxis occurs secondary to capillary fragility, rarely is there an increase in the bleeding time.

The pregnant patient with osteogenesis imperfecta presents a special problem, that is, her fetus may also be affected by the disease. Both the obstetrician and the anesthetist, therefore, must be knowledgeable about this disease. Identification of the fetus with osteogenesis imperfecta congenita by abdominal x-ray during the last few weeks of pregnancy (which will show fetal fractures, and generally, poor calcification) will lead to a heightened awareness of the child's fragility as a neonate. This will decrease both neonatal morbidity and mortality.

The patient with osteogenesis imperfecta presents complex problems to the anesthetist. The pathophysiology of the disease process must be understood to enable successful management of...
the case. While Dunham and Spellacy\textsuperscript{8} feel that there may be decreased fertility in patients with osteogenesis imperfecta, seven cases have been successfully managed in this obstetrical unit (University of Cincinnati Medical Center) during the past two years. As all these young women have the potential for 20 or more child-bearing years, the possibility that the anesthetist will encounter a patient with osteogenesis imperfecta is increasing.

**The case**

A 13-year-old black female with osteogenesis imperfecta congenita was followed to term and scheduled for elective cesarean section. The obstetrical decision to perform a surgical delivery was predicated on borderline cephalopelvic measurements, and the concern for fracture of the pelvis if vaginal delivery was attempted. An abdominal x-ray revealed a singleton fetus with no evidence of skeletal fractures. The abdominal x-ray was performed because in the congenita variant of osteogenesis imperfecta, the fetus may sustain intrauterine fractures and may die before or during attempted vaginal delivery.\textsuperscript{8} This family's history of osteogenesis imperfecta included the patient's mother and three of her six children.

Past medical history revealed several childhood fractures, including both wrists and a femoral fracture sustained when she was turned in her crib as an infant. Current laboratory data revealed normal platelet function, clotting parameters, electrolytes, urinalysis and complete blood count. Physical deformity of the thorax was not evident, and respiratory function was judged to be within normal limits. Dentition, however, was poor.

A general anesthetic technique was chosen on the basis of the patient's labile teenage personality and the possibility of a vertebral fracture from the needle placement. (At least one author, however, has reported successful use of epidural anesthesia.\textsuperscript{9}) After administration of an oral antacid, the patient was carefully transferred to the operating room table. Particular care was given to positioning and padding of the extremities and bony prominences. Her arm was padded before applying the blood pressure cuff, as fractures of the humerus have been reported from this procedure.\textsuperscript{4}

Atropine 0.6 mg, and d-Tubocurarine 18 mg, were given intravenously following 10 minutes of preoxygenation. Induction was accomplished with 250 mg thiopentone administered intravenously. Muscle relaxation was not maintained with succinylcholine because Soloman\textsuperscript{9} and others have reported hyperthermia in patients with osteogenesis imperfecta under general anesthesia. A second consideration in the use of succinylcholine would be the potential for fractures occurring with major muscle fasciculation.

Temperature monitoring was performed with an esophageal probe throughout this surgical procedure, with the patient's temperature remaining within normal limits throughout the procedure and postoperative period. Endotracheal intubation was accomplished with gentle movement of the mandible and minimal extension of the neck to avoid fractures. Sellick's maneuver was maintained to prevent aspiration of the gastric contents. The teeth were not damaged.

Anesthesia was maintained with nitrous oxide-oxygen in a 50:50 mixture until delivery of the infant. Following delivery, morphine sulfate 10 mg, was given in divided doses and the nitrous oxide was increased to 60\% to maintain anesthesia. The patient was extubated when awake. She had intact reflexes present at the end of the case. The term male infant weighted 6 lbs. 15 oz; the Apgar scoring was 5 and 8 at 1 and 5 minutes, respectively.

The postoperative course was complicated by a low-grade fever of unknown etiology. Blood and urine cultures were negative. Radiologic studies failed to reveal the presence of fractures or the suggestion of osteomyelitis. The patient was discharged to her home in good condition on the tenth postoperative day. At last report, the infant showed no evidence of osteogenesis imperfecta.

**REFERENCES**


**AUTHORS**

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