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Case Report

The role of the multidisciplinary team in managing a challenging brittle bone disease with 18 fractures in the modern era: a case report and literature review

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ABSTRACT

Osteogenesis imperfecta (OI) is a rare autosomal dominant disorder due to collagen type I biosynthesis defects. It aggressively affects maternal and fetal outcomes in pregnancy. In this case report, we will be discussing the course of pregnancy in an OI patient who has already had 18 fractures since childhood and the challenges faced during the course. A 29-year-old primigravida who was diagnosed with OI since childhood had approximately 18 fractures up to the age of 29. She was under regular antenatal follow-up in our clinic. She was diagnosed with late-onset stage 1 fetal growth restriction at 36 weeks. She was planned for an elective caesarian under a multidisciplinary team at 37 weeks given her short stature, contracted pelvis, and stage 1 fetal growth restriction. She had an uneventful delivery course without any intraoperative and postoperative complications. There was no stress fracture post-delivery, and the newborn developed normally during a 1-year follow-up period. This case highlights the management of OI in pregnancy who had a history of multiple fractures and also emphasizes the need for a tertiary care center in the management of these high-risk patients. A close follow-up of these patients with prenatal genetic testing, regular ultrasound monitoring, and a multi-disciplinary team will aid in an early diagnosis of all the potential underlying complications, thereby leading to favorable maternal and fetal outcomes. A literature review on this topic is also presented.

Keywords: Cesarean section, Osteogenesis imperfecta, Pregnancy, Case report

INTRODUCTION

Osteogenesis imperfecta (OI) also known as brittle bone disease is a rare, autosomal dominant disorder caused by mutation in the genes encoding alpha-1 and alpha-2 chains of type I collagen. Although rare with an incidence of around 1 in 20, 000 live births, it is still the most common inherited disorder of connective tissue.[1] Patients present with multiple repetitive fractures leading to skeletal deformities, short stature, scoliosis, laxity of skin and ligaments, hearing loss, blue sclera, dentinogenesis imperfecta, platelet dysfunction, and heart disease.[2]

OI also poses significant maternal and fetal complications which pose a challenge in delivering in these patients, with few reported cases of increase in cesarean sections, preeclampsia, premature rupture of membranes, preterm delivery, placental abruption, and vaginal tears.[3] It is therefore pertinent to offer multi-disciplinary care for patients with OI to assess and discuss the implications of OI in pregnancy and delivery of the fetus. Limited information has been discussed in the literature regarding the management of OI in the intrapartum period and also serial ultrasound monitoring, a proper protocol has still been not established for the same

CASE REPORT

Our patient was a 29-year-old primigravida who presented to our antenatal clinic at 20 weeks period of gestation, she was under follow-up in our Orthopedics department given

OI. For this case, ethical approval and patient consent have been sought. At the age of five, she started to have fractures after trivial injuries, she was diagnosed with OI. She had multiple fractures 18 times, and she was wheelchair-bound by 17 years of age. A corrective osteotomy with Ilizarov's applicator of the femur was done then she became ambulatory with the prosthesis. Since then, she was not on any medications other than calcium tablets on and off. She had a regular menstrual cycle. She is a non-smoker and abstains from alcohol. She had no family history of OI and no history of short stature. She was booked with us for 20 20-week period of gestation and was on regular follow-up in our antenatal clinic. She was on iron and calcium tablets during the antenatal period.

On clinical examination, her height was 122 centimeters Figure 1 with right shoe orthosis, her weight was 42 kilograms, and has a body mass index of 28.2. She had a blue sclera (Figure 2) with a triangular face. There was no pallor, icterus, cyanosis, clubbing, or pedal edema. The dentition was poor. She had a lynching gait with scoliosis and marked deformities in bilateral lower limbs. The right leg was shortened with anterior bowing of the tibia and the left leg arch was deformed. Bilateral knee joint flexion was normal. The straight leg raising test was up to 90 degrees. The hip joint was contracted. Breast and thyroid examinations were normal. Cardiovascular and respiratory findings were normal without any abnormalities. Abdomen examination revealed a 34-week size uterus at 37 weeks period of gestation, with a fetal heart rate of 142 beats per minute, cephalic presentation, longitudinal lie, and relaxed uterus. Her antenatal period was uneventful. Given the complexity of the case, A multidisciplinary team (MDT) team consisting of a panel of obstetricians, a pulmonary medicine team, cardiologists, neonatologists, anesthetists, maternal-fetal medicine experts, and a genetics team met to discuss the challenges of ongoing pregnancy in this complicated patient. The patient was counseled by our genetics team regarding the risks of OI in the fetus and the risk of gross congenital anomaly in the fetus since aneuploidy screening was not done due to her late follow-up in our clinic. A detailed anomaly scan was performed at 20 weeks of gestation and revealed no signs of OI in the fetus although the patient was counseled regarding mild forms not being evident on an ultrasound. Her blood group was B positive; she had mild anemia. HPLC showed borderline low HbA2. Viral markers were nonreactive. Liver and kidney function tests were normal. 25 hydroxylase levels were 13.6 ng/ml. ECG was of normal sinus rhythm. The chest and spine radiographs showed kyphoscoliosis (Figure 3). Maternal echo-cardiogram showed mild anterior mitral valve prolapse and mild mitral regurgitation with normal left ventricular ejection fraction. Serial growth scans were performed at two weekly intervals given the high-risk nature of the disease and reported growth restriction; growth was normal till 34, 34-week period of gestation. But ultrasound of fetal growth at 36 weeks period of gestation revealed symmetric stage 1 fetal growth restriction, Umbilical artery, and middle cerebral artery dopplers were in the normal range. This finally culminated in an elective caesarian section at 37 weeks period of gestation given OI with short stature and contracted pelvis.

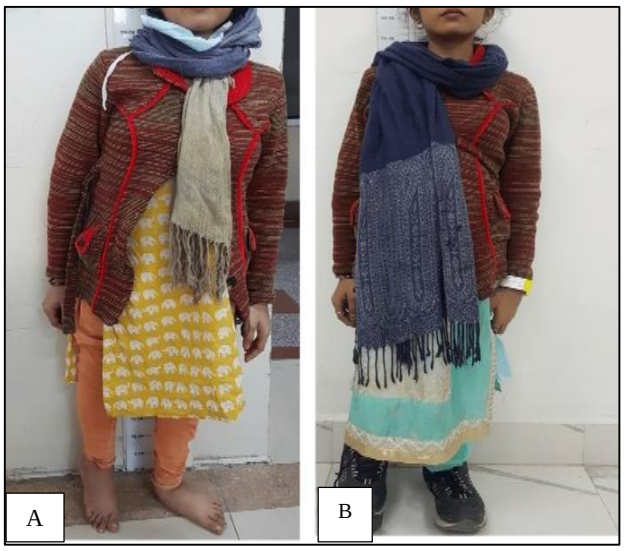

Figure 1 (A and B): Short stature of the patient.

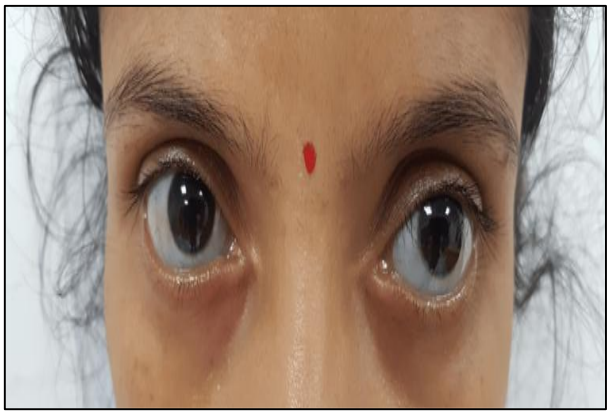

Figure 2: Eye depicting blue sclera.

Spinal anesthesia with bupivacaine was administered to avoid the potential risks associated with general anesthesia. Before attempting regional anesthesia, the airway and possibility of difficult intubation were assessed with direct laryngoscopy under topical anesthesia of the oropharynx, in case there was regional block failure. To secure the neutral positioning of the limbs and support the angulation of the pelvis, foam padding was used. A male baby was delivered, weighing 2305 grams with an APGAR score of 8 and 9 after 1 and 5 min, respectively. In post postpartum period, contraceptive counseling was done and she was given depot medroxy progesterone acetate injection. The neonate was monitored keenly for any evidence of OI. There were no signs of respiratory distress, fractures at birth, hearing or vision defects. The baby was under regular follow-up, The developmental milestones were normal and no fractures have been reported yet.

DISCUSSION

This case highlights the difficulties and challenges faced in the management of OI in pregnancy. Finally, our successful management of this case identifies how prenatal ultrasound monitoring and regular antenatal follow-up are necessary to provide favorable maternal and fetal outcomes and also how a multidisciplinary approach is needed to provide such treatment. This case highlights the paradigm of the challenges faced in the antenatal care of a patient with OI in pregnancy.

OI, is an autosomal-dominant disorder caused by mutation in the genes encoding alpha-1 and alpha-2 chains of type I collagen.[2] There are up to 15 subtypes of OI depending on the clinical and radiological manifestations along with the underlying genetic cause.[4] It is characterized by multiple fractures and other abnormalities including short stature, scoliosis, basilar skull deformities, blue sclera, hearing loss, dentinogenesis imperfect, laxity of the ligaments and skin, platelet dysfunction, and heart disease.[1] Type I is the most common and mildest form with a variable fracture rate, minimal deformity, normal stature, and hearing impairment.[5]

Fertility is preserved, especially in those patients with type I of the disease, and pregnancy can be carried to term. Pregnant women affected by type I OI should be closely monitored to assess fetal well-being and detect pregnancy-related complications associated with an increased risk for osteoporosis, restrictive pulmonary disease, other problems related to connective tissue disorders, cephalopelvic disproportion antepartum hemorrhage, pre-eclampsia, placenta abruption, vaginal tears, premature rupture of membranes, preterm birth, intrauterine growth restriction. Fetal adverse events include small-for-gestational-age infant and the congenital malformation.[3]

Stress fractures are one of the defining features of OI. It can occur in both vaginal and abdominal deliveries.[6,7] In the case of our patient, the only complication faced was stage one fetal growth restriction, which was identified by the serial growth scans and was managed timely, we had no other complications in the managing the mother and baby.

Ideally, genetic counseling is sought before conception. Once pregnant, prenatal diagnosis can be established by chorion villous sampling and amniocentesis. Serial scans would identify the affected fetus with fractures.[8] The prenatal sonographic findings are short bowed femurs with fractures, deformity of the long bones, and ribs, thin skull with unusual clarity of intracranial structures, multiple fractures, and reduced echogenicity.[9] Though prenatal diagnostic testing was not done on our patient, prenatal ultrasound did not show any evidence of fractures or deformities.

It has been shown that prenatal diagnosis has no influence on the mode of delivery in most of the cases.[10] And the also mode of delivery remains controversial and should be determined on an individual basis.[3] Most of the cases reported have proposed cesarean section as the ideal route of delivery in OI patients due to pelvic deformities, cephalopelvic disproportion, and increased incidence of abnormal fetal presentation.[6,7,11] It was also shown that cesarean section neither decreased the rate of fractures at birth in infants with nonlethal OI nor did it prolong survival for those with lethal forms.[10] In a particular study, a cesarean section rate in OI patients was 54% and the most common indication was a non-vertex delivery at 53%, and an antenatal diagnosis of OI was made in less than 15%, the rate of breech presentation at term was 37%. But strikingly a significant association was found between the mode of delivery and the OI subtypes, and also increased rate of cesarean sections was found in nulliparous patients and in patients undergoing prenatal genetic counseling.[10]

Anesthesia in patients with OI poses difficulties like difficulty in the insertion of regional anesthesia, difficult airway control, and respiratory insufficiency due to the deformed thoracic cavity. Airway management can be particularly challenging due to brittle teeth, weak cervical vertebrae, and mandible, increased risk for odonto-axial dislocation, and difficult airway.[12] A higher risk of perioperative hyperthermia has also been reported.[13] This case exemplifies the successful use of regional anesthesia as described in other patients with OI in literature, which decreased the need for endotracheal intubation and its associated risks of aspiration and bone injury. Regional anesthesia poses challenges of its own due to severe kyphoscoliosis, coagulopathy, and inability to maintain the supine position awake.

Yimgang et al reported the neonatal outcomes in neonates born to OI patients, of the babies who had premature birth, 31% were requiring neonatal intensive care unit (NICU) admission. Bone deformities were noted in 18% and fractures in 18% of neonates, while 22% had respiratory complications.[14] In our patient, there was no unfavorable outcome noted in the baby in the year follow-up period.

CONCLUSION

We present a unique case of pregnancy in an OI patient, the management of the challenges faced in this case including antepartum, intrapartum, and postpartum period. This case also highlights the importance of close antenatal follow-up of these patients in a tertiary care center guided by a multidisciplinary team to provide prompt management and identification of complications that can arise secondary to this. Because of the diverse nature of the disease and its associated life-threatening risks to the mother and the fetus, we have also identified that further research is needed into the management of this condition, specifically regarding ultrasound monitoring and intraoperative management protocols.

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