OSTEOGENESIS IMPERFECTA IN A NORMAL PREGNANCY : A CLINICAL CASE REPORT

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ABSTRACT

The aim of this work is to report a clinical case of childbirth in a patient with Osteogenesis Imperfecta (OI) during an unplanned pregnancy. Pregnancy in these patients is associated with maternal and fetal complications, given that the presence of an autosomal dominant mutation in the mother determines a 50% risk of OI in the offspring. This is a case report of a pregnant patient with Osteogenesis Imperfecta, with a positive maternal and fetal outcome, aiming to discuss possible complications arising during pregnancy due to this comorbidity, focusing on recommendations for managing these women. Patient C.C.V., a 25-year-old female, diagnosed with OI, primiparous, with a history of multiple bone fractures, bone dysplasia, blue sclera, and endometrioma. She was admitted in March, 2023, for a cesarean section, hemodynamically stable with unaltered laboratory tests and negative serologies, with a family history of diabetes, heart disease, and glaucoma. The patient reported an unexpected pregnancy without prenatal care, and the fetus showed no suspicion of pathology. The delivery was via cesarean section, after the onset of labor, with spinal anesthesia in the operating room of the maternity hospital. The fatus was born healthy, without complications, and is not affected by OI. This case highlights the importance of multidisciplinary care and planning for pregnancies in women with OI, as well as the significance of comprehensive knowledge of the patient's history of previous fractures and clinical conditions, so that obstetricians and anesthesiologists can assist in choosing the best delivery method for both the mother's and the fetus's health.

KEYWORDS: CONGENITAL BONE DISEASES, PREGNANCY, OSTEOGENESIS IMPERFECTA, PRENATAL.

INTRODUCTION

Osteogenesis Imperfecta (OI), also known as brittle bone disease, is a group of connective tissue disorders with heterogeneous presentation, related to a deficiency in the synthesis of type I collagen. Its incidence is estimated at 1 case per 15,000-20,000 births, making it the most common genetically inherited connective tissue disorder[1].

Of these cases, 85 to 90% are caused by autosomal dominant structural or quantitative mutations in collagen-related genes, with emphasis on the COL1A1 and COL1A2 genes, which encode the alpha chains of type 1 collagen. The location of this mutation within the protein determines the genotypic and phenotypic presentation of patients with OI[2].

In 1979, a classification of OI subtypes was developed based on clinical characteristics and severity. Type I is mild, non-deforming, with blue sclera; type II is characterized by a perinatal lethal form; type III is severe, with progressive deformity; type IV is moderate, with normal sclera[3]. Regarding genetic characteristics, 22 distinct types of the disease have been identified.

In general, clinically, this pathology is characterized by bone fragility and deformities, associated with fractures from minimal trauma. It can also cause growth deficits depending on the clinical form presented by the patient. As it is a disorder of connective tissue, findings can extend to other systems besides the skeletal system, such as blue sclera, hearing loss, dentinogenesis imperfecta, and pulmonary dysfunction[1].

With advances in the management of OI, there has been an increase in the life expectancy of patients, and more women affected by the disease reach reproductive age with a desire for pregnancy. Pregnancy in these patients is associated with maternal and fetal complications, as the presence of an autosomal dominant mutation in the mother determines a 50% risk of OI in the offspring[4]. Therefore, reproductive counseling in this group is essential, as well as proper management in prenatal care and childbirth to minimize negative outcomes among pregnant patients.

This is a case report of a patient with Osteogenesis Imperfecta (OI) who became pregnant, with a positive maternal and fetal outcome. The aim is to discuss possible complications of pregnancy due to this comorbidity, focusing on management recommendations for these women.

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CASE REPORT

Patient C.C.V., 25 years old, was admitted in March 2023 for cesarean section, hemodynamically stable, with normal laboratory tests and negative serologies. Primiparous, she became pregnant naturally, without planning, and was 36 weeks pregnant. She discovered the pregnancy during an ultrasound exam for preoperative evaluation before the removal of an endometrioma in the left ovary. The patient did not have prenatal care. She has osteogenesis imperfecta (type not specified), diagnosed at 6 months of age, with a history of more than 20 fractures, blue sclera, and a family history of genetic diseases such as heart disease, glaucoma, and diabetes. Despite the osteogenesis imperfecta, she has no other comorbidities, does not take medications, and has no history of hypertension, diabetes, or smoking. However, she reports social alcohol consumption. A karyotype was not performed. The fetus did not show any suspected pathology.

An elective cesarean section was performed with spinal anesthesia, a regional anesthesia with the injection of anesthetic into the subarachnoid space, blocking pain in the lower body. After delivery, a living child without OI, without apparent injuries, and with adequate vital signs was born.

DISCUSSION

Osteogenesis imperfecta, a rare and hereditary condition of the connective tissue, presents significant challenges for affected individuals and their families. Characterized by bone fragility and susceptibility to fractures due to defects in collagen synthesis, this condition requires a careful approach, especially during pregnancy.

For women planning to conceive, it is advisable to seek genetic counseling before conception in cases of osteogenesis imperfecta. This can help families better understand the implications of the condition and make decisions about family planning.

During pregnancy, meticulous preparation is essential, allowing for the monitoring of fetal development, early identification of complications, and guidance on appropriate prenatal practices. Complications during pregnancy, which can range from intense pain and bone deformities to more serious situations such as uterine rupture, highlight the importance of prenatal monitoring. This process requires a multidisciplinary approach, involving professionals such as obstetricians, geneticists, and psychologists, who should be fully aware of the patient's life history, including her fractures and complications resulting from the disease, to provide accurate and effective guidance during the preparation for pregnancy, childbirth, and the postpartum period.

So far, there is no definitive drug treatment for this disease. However, several methods have been associated with monitoring affected patients. One such approach includes the use of bisphosphonates, investigated as an alternative to improve bone density and reduce the chances of fractures in patients diagnosed with OI. However, the lack of conclusive scientific studies supporting the beneficial efficacy of these drugs during pregnancy highlights uncertainty about their safety during the gestational period.

Precautions during childbirth are expanded to ensure the health of all involved. The team must be prepared to offer support and specific interventions for each patient, aiming for a unique experience and adapting to the specific needs of each pregnant woman with OI, such as additional care with anesthesia, intubation, padding of the stretcher, and equipment used, among others.

The administration of anesthesia can generate uncertainty, especially when choosing medications, and challenges during tracheal intubation. It is recommended to apply anesthetics to the oropharynx area before laryngoscopy, while the patient is conscious, to assess possible difficulties in intubation. Furthermore, intubation via bronchofibroscopy and the use of a laryngeal mask airway are alternatives that should be considered[10].

Delivery, often surgical, may be necessary due to the pregnant woman's pelvic deformities, cephalopelvic disproportion or the fetus' condition with osteogenesis imperfecta10. These considerations highlight the complexity of the act in patients with this condition and the importance of specific care to ensure safe and positive results.

Due to reports of different techniques, it is essential to analyze each case individually to determine the best approach to factors such as anesthesia and mode of delivery, a procedure implemented in the present clinical case. One study found that hemorrhage is a possible complication in 10% to 30% of patients with OI, due to tissue fragility and inadequate response to bleeding caused by collagen deficiency, which increases the risk of postpartum uterine atony[7]. During childbirth, it is crucial to observe special care, ensuring, for example, adequate positioning to avoid compression. Additionally, research has shown high rates of obstetric complications in women with osteogenesis imperfecta, including gestational diabetes, cesarean section, need for blood transfusion and occurrence of fractures both before and after childbirth[8].

Diagnosis of fetal status can be conducted through the extraction of chorionic villi and imaging observations of the fetus[9]. During pregnancy, an ultrasound can detect OI and indicate the type, as the most serious and potentially fatal form of osteogenesis imperfecta. After the birth, the doctor checks the symptoms and performs a physical examination to make a diagnosis. If any doubts persist, the professional may choose to perform a skin biopsy or take a blood sample for genetic analysis, methods that help confirm the diagnosis and define the most appropriate treatment for the condition.

CONCLUSION

It is ideal that the pregnancy for those with Osteogenesis Imperfecta is planned and monitored by professionals who are experts in the patient's clinical condition and have extensive knowledge of the specific case to avoid future complications during pregnancy and childbirth. In the case reported, the patient had an unplanned pregnancy but was well managed and monitored, without serious complications.

After the pregnancy is discovered, the patient with OI must receive a multidisciplinary approach, involving professionals such as obstetricians, geneticists and psychologists, who are aware of the patient's life history, including her fractures and complications resulting from the disease. There are specific risks related to this condition during pregnancy, especially related to hemorrhages, due to tissue fragility and inadequate response to bleeding caused by collagen deficiency, which increases the risk of postpartum uterine atony, in addition to an increased risk of obstetric complications.

To date, there are no medications proven to be effective for treating this condition and there is a risk that the fetus may also be a carrier of this genetic condition. It is important and necessary to monitor and identify early OI in the baby during pregnancy.

It is generally recommended that the birth be cesarean section, given the cephalopelvic disproportion and/or pelvic deformations resulting from previous fractures. A careful analysis by the anesthetist and obstetrician is important to assess the best route of delivery for both the mother and the baby.

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