CASE REPORT

Successful obstetric and anaesthetic management of a pregnant woman with achondroplasia

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SUMMARY

Achondroplasia is the most prevalent form of dwarfism, and there is little evidence about the optimal management of pregnant women with achondroplasia. We presented a 25-year-old primigravid woman with achondroplasia who was followed up during the pregnancy period and performed elective caesarean section with combined spinal-epidural anaesthesia at the 38th week of gestation. Frequent obstetric follow-up visits and invasive prenatal diagnostic tests should be offered during the antenatal period due to the increased risk for obstetric complications, such as premature delivery and fetal anomalies. Prenatal detailed counselling, comprehensive evaluation of the potential risks, obstetric and perioperative management should be performed by a multidisciplinary care team, including an obstetrician, anaesthesiologist, pulmonologist, cardiologist and neonatologist.

BACKGROUND

Achondroplasia is an autosomal-dominant disorder and is the most prevalent form of dwarfism with an incidence of 1 in 15 000.[1] Despite the autosomal-dominant transition, >80% of cases occur due to a spontaneous mutation.[2] The fertility rate of women with achondroplasia is usually low[3]; thus, there is little evidence about the optimal management of pregnant women with achondroplasia. The potential problems of pregnancy in a woman with achondroplasia include increased caesarean delivery rate, higher preterm birth risk and fetal skeletal dysplasia incidence. Besides, cardiac abnormalities including congenital heart diseases, cardiomyopathy, coronary artery disease and pulmonary hypertension are increased in achondroplastic pregnant patients. Also, pulmonary complications including recurrent respiratory infections, upper airway obstruction, scoliosis, rib deformities and restrictive lung diseases are common in these pregnancies.[4] Furthermore, anaesthetic management of pregnant women with achondroplasia is challenging because general and regional anaesthetic techniques can pose several problems for these patients. This case presentation describes the obstetric and anaesthetic management of a pregnant woman with achondroplasia who presented to our clinic.

CASE PRESENTATION

Presentation

A 25-year-old primigravida woman with achondroplasia was admitted as a high-risk pregnancy at the outpatient clinic for routine obstetric care in the gestational week 9. Her medical history was unremarkable. She was 106 cm tall and weighed 53 kg with a body mass index of 47.1 kg/m^2. Her partner was also achondroplastic. She was referred to the departments of anaesthesiology, pulmonary medicine and cardiology.

Investigations

She had short extremities with a normal size head, and she had no neurological or musculoskeletal symptoms. An examination revealed a Mallampati II airway with a 4 cm thyromandibular distance, allowing comfortable breathing and no severe kyphoscoliotic deformity, was observed in the thoracolumbar spine. Baseline pulmonary function testing revealed no pathology: the forced expiratory volume in 1 s/forced vital capacity ratio was 89%, the forced expiratory volume in 1 s was 126% and the forced expiratory flow was 25%–75% of predicted. Transthoracic echocardiography demonstrated normal biventricular systolic and diastolic functions, minimal tricuspid regurgitation and no evidence of pulmonary hypertension. Although the first trimester-combined test revealed low trisomy 21 and trisomy 13/18 risk (1/3745 and <1/10 000, respectively), the patient and her partner were informed about the risk of fetal skeletal dysplasia and were offered a prenatal diagnosis. Due to a desire to continue the pregnancy regardless of the outcome, they refused an invasive prenatal diagnosis. A routine mid-trimester fetal ultrasound scan revealed normal fetal anatomy. Biometric measurements of fetal long bones were <5th percentile in week 28 of gestation. No pathological findings were detected during routine obstetric follow-up visits until week 38 of gestation.

Management

An elective caesarean section was planned due to a cephalopelvic disproportion (CPD) detected during week 38. She was preoperatively counselled about anaesthetic difficulties, the possible need for general anaesthesia and the requirement for postoperative intensive care. Baseline pulmonary function test and transthoracic echocardiography were repeated and the results were detected similar to the results at the first trimester of pregnancy. During the preoperative anaesthetic evaluation, only access to only one peripheral vein was obtained from the right antecubital fossa. We were only able to access the intervertebral space with the aid of

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ultrasonographic guidance. Preparation of the equipment for the central venous catheter replacement was carried out considering the possibility of the excessive bleeding but no more intravenous access was required during the surgery. We performed combined spinal–epidural anaesthesia, even after the technical difficulties due to the spinal abnormalities. An 18-gauge Tuohy needle was inserted into spinal space at the level of L3–L4 interspace. In total 1,2 mL containing a mixture of fentanyl 10 µg and 0.5% hyperbaric bupivacaine 5 mg were administered into the spinal space. Then, the epidural catheter was inserted. Maternal heart rate and arterial pressure were well controlled during the procedure. Caesarean section was started 10 min after the performance of the neuraxial anaesthesia. A 49 cm female was delivered after a Pfannenstiel skin incision. The neonate weighed 2640 g and had APGAR scores of 9 and 10. The diagnosis of achondroplasia was confirmed by a neonatologist according to the findings of physical examination, radiographic evaluation and karyotype analysis that showed the presence of fibroblast growth factor receptor 3 mutation. No complications were detected at the maternal and neonatal postoperative follow-up visits, and the mother and neonate were discharged from the hospital 3 days after the surgery. The puerperal period was uneventful for the mother. At the time of writing, the baby showed normal growth according to the specialised growth curves developed for the children with achondroplasia and no additional intervention was planned by the Paediatric Orthopaedic Clinic.

DISCUSSION

Caesarean section is the usual mode of delivery for women with achondroplasia due to the CPD. The contracted pelvis of the mother and a restricted foramen magnum and cervical spine of the neonate are responsible for these requirements, and the fetus may have an increased risk for spinal cord compression during manipulation of the neck in the second stage of delivery.[5] Therefore, a vaginal delivery may be dangerous, so an elective caesarean section is preferable in these cases. The presence of a neonatologist in the delivery room is beneficial because of the increased risk of respiratory compromise in the immediate newborn period and the potential difficulty with neonatal resuscitation. Also, there is increased risk for fetal distress due to the enhanced likelihood of hydrops fetalis, severe polyhydramnios and fetal visceral abnormalities in these pregnancies.[6 7] Ayoubi *et al* reported that the timing of the delivery should be determined by balancing the risks of a preterm birth with the mother's respiratory problems, and they reported a case of diastrophic dwarf pregnant women who had an elective caesarean section planned for week 32 of pregnancy due to maternal respiratory compromise.[8] Lagoy *et al* presented a study about a pregnant woman with diastrophic dysplasia who underwent caesarean section at week 35 of pregnancy with a midline incision for the purpose of better visualisation of the lower abdomen and lower uterine segment. In this case, an elective caesarean section was planned in week 38 of gestation with a Pfannenstiel skin incision due to CPD.[9] A neonatologist was available in the delivery room, and the neonate did not require any resuscitation effort.

A significant decrease in functional residual capacity and an increment in closing volume secondary to restrictive lung disease may cause atelectasis and ventilation/perfusion problems in achondroplastic pregnant patients. In the achondroplastic pregnant patient, cardiology and pulmonology consultations were performed at the beginning of the pregnancy, and there was no pathological finding in her pulmonary function test and echocardiography except minimal tricuspid regurgitation.

Homozygous achondroplasia is a condition with severe skeletal deformities that is lethal within the first months of life. There is a 25% risk of conceiving a child with homozygous achondroplasia when both parents have achondroplasia.[10] Prenatal diagnostic procedures for the detection of homozygous achondroplasia can be performed in the first or second trimester of pregnancy due to shortening of long bones can become evident only in the third trimester of pregnancy.[11] For this reason, we recommended invasive prenatal diagnosis in the first trimester of pregnancy for the early detection of homozygous achondroplasia by DNA-based methods on chorionic villi.

The technical difficulties of local anaesthesia due to lumbar lordosis, thoracic scoliosis, poor landmarks, spinal stenosis, engorged epidural veins and narrowed epidural and intrathecal spaces may cause a dural puncture or catheter placement failure and increase the epidural/spinal block level.[12] On the other hand, performing general anaesthesia can result in severe difficulties due to a large head and tongue, restricted ability to extend the head, instability of the cervical spine and narrowed upper airway.[13 14] The anaesthetist must consider the benefits and limitations of each method for these patients on an individual basis. In this case, we administered combined spinal–epidural anaesthesia under ultrasonographic guidance and did not encounter any anaesthetic problems during the preoperative and postoperative periods with close monitoring and dose adjustments.

Learning points

- Obstetric and perioperative management should be performed by a specialist multidisciplinary care team comprising an obstetrician, anaesthesiologist, pulmonologist, cardiologist and neonatologist.
- Invasive prenatal genetic testing should be offered.
- Frequent obstetric visits with regular ultrasound evaluation should be performed to detect pregnancy complications and monitor fetal long bone growth.
- Maternal respiratory and cardiac functions should be checked regularly during the pregnancy due to the increased maternal morbidity and mortality associated with pulmonary and cardiac disease and increased maternal respiratory compromise, particularly during the late gestational period.
- Anaesthetic assessment should be carried out early in the third trimester because of the anaesthetic management challenges and the risk of an unscheduled emergency caesarean.

Contributors RM: conception or design of the work. SE: data collection. EC: critical revision of the article.

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REFERENCES

1 Li X, Duan H, Zuo M. Case report: Anesthesia management for emergency cesarean section in a patient with dwarfism. *BMC Anesthesiol* 2015;15:59.
2 Huang J, Babins N. Anesthesia for cesarean delivery in an achondroplastic dwarf: a case report. *Aana J* 2008;76:435–6.
3 Ansari MH, Abraham A. Anaesthetic management of unexpected subglottic stenosis in an achondroplasic dwarf. *Acta Anaesthesiol Scand* 2004;48:928–9.

4 Berkowitz ID, Raja SN, Bender KS, *et al*. Dwarfs: pathophysiology and anesthetic implications. *Anesthesiology* 1990;73:739–59.
5 Thomas IT, Frias JL, Williams JL, *et al*. Magnetic resonance imaging in the assessment of medullary compression in achondroplasia. *Am J Dis Child* 1988;142:989–92.
6 DeRenzo JS, Vallejo MC, Ramanathan S. Failed regional anesthesia with reduced spinal bupivacaine dosage in a parturient with achondroplasia presenting for urgent cesarean section. *Int J Obstet Anesth* 2005;14:175–8.
7 Krakow D, Lachman RS, Rimoin DL. Guidelines for the prenatal diagnosis of fetal skeletal dysplasias. *Genet Med* 2009;11:127–33.
8 Ayoubi JM, Jouk PS, Pons JC. Diastrophic dwarfism and pregnancy. *Lancet* 2001;358:1778.
9 Lagoy JS, Kofford ND, Gosselin BJ, *et al*. Management of a parturient with diastrophic dysplasia. *A A Case Rep* 2015;5:6–8.
10 Gooding HC, Boehm K, Thompson RE, *et al*. Issues surrounding prenatal genetic testing for achondroplasia. *Prenat Diagn* 2002;22:933–40.
11 Rani R, Singh S, Singh S, *et al*. Term Pregnancy in an Achondroplastic Dwarf: A Case Report. *Indian Journal of Clinical Practice* 2014;25:364–6.
12 Kumar MM, Forster MR. Combined spinal epidural anaesthesia for elective caesarean section in a patient with spondylometaphyseal dysplasia. *Int J Obstet Anesth* 2002;11:225–7.
13 Mitra S, Dey N, Gomber KK. Emergency cesarean section in a patient with achondroplasia: An anesthetic dilemma. *J Anesth Clin Pharmacology* 2007;23:315–8.
14 Porter M, Mendonca C. Anaesthesia for Caesarean section in a patient with diastrophic dwarfism. *Int J Obstet Anesth* 2007;16:145–8.