

Successful pregnancy and delivery in a woman with achondroplasia

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Abstract

Achondroplasia is a common form of dwarfism occurring in one out of 25,000 of live births. These patients present many problems during pregnancy and at the time of the delivery. The anaesthetist may also face several difficulties for both general and regional anaesthesia. The aim of this case report is to discuss various obstetric and anaesthetic considerations in such patients and ways to manage the difficulties. A 29-year-old woman expecting her first child, with achondroplasia —height of just 3'2" (98 centimetres) — at 31 weeks of gestation, presented to our OPD for antenatal visit on May 15, 2018. Her successful elective lower segment caesarean section was performed on June 25, 2018 at 37 weeks in view of contracted pelvis under general anaesthesia. A live male baby was delivered, with no complications, good Apgar score and birth weight of 2.7kg. This shows that a woman with a genetic disorder like achondroplasia can become pregnant and deliver a healthy child at term.

Keywords: Achondroplasia, pregnancy, Kyphoscoliosis, infertility.

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Introduction

Achondroplasia is a genetic illness that causes disproportionate dwarfism, patients having short limbs as compared to trunk and are short in height.¹

Achondroplasia is caused by mutations in the Fibroblast Growth Factor Receptor 3 gene. There are two sites of mutations in the FGFR3 gene which are responsible for all cases of achondroplasia.² These mutations cause the FGFR3 protein to be excessively active, which restricts the skeletal development and leads to the abnormal bone growth. Inheritance of achondroplasia is autosomal dominant. In most cases (about 80 percent) the problem is not inherited and have average size parents, these cases results from new mutation in FGFR3 gene, while remaining cases have inherited altered gene from one or two affected parent. If one parent is affected the child has 50 percent chance of acquiring the disease and 50 percent chance to be the

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normal. If both parents are affected, than there is 25 percent chance of having child with average stature, 50 percent chance of having heterozygous achondroplasia (one defective gene) and 25 percent chance for homozygous achondroplasia (two defective genes) which is the fatal type.^{1,2}

Common diseases seen in these patients are infertility, menstrual irregularities, dysmenorrhoea, leiomyoma and premature menopause.³

Conception is unusual for a dwarf and if she becomes pregnant then there is a great risk of early pregnancy loss because of the small size of the uterus. Obstetric problems, such as polyhydramnios, pre-eclampsia, premature delivery and contracted pelvis may occur in these patients. Foetal complications, such as hydrocephalus, thoracic cage abnormalities and foetal demise, may occur. The foetus may also present with achondroplastic features because affected heterozygous patients have 50% risk of transmitting the disease to their offspring. Mode of delivery should be a caesarean section because the size and shape of the pelvis causes cephalopelvic disproportion.⁴

The choice of anaesthesia —whether general or regional— depends upon individual cases, and the anaesthetist may confront many problems during the latter stages of pregnancy.⁵ There may be difficulty in tracheal intubation during general anaesthesia due to foramen magnum stenosis and instability of cervical spine. Cervical cord compression may occur due to over extended neck, so it must be avoided. A number of anatomical deformities in such patients may complicate the techniques of regional anaesthesia. Kyphoscoliosis, narrowing of the vertebral canal, can reduce the distance between the pedicles and osteophyte which can produce difficulty for spinal anaesthesia.⁶

There are no definite recommendations regarding anaesthetic procedures during pregnancy. The decision has to be taken on individual basis after a detailed risk-benefit analysis.⁵

We describe a case of a patient with achondroplasia who had a successful delivery through caesarean section.

Case Report

A 29-year-old primigravida, only 98 centimetres tall and married for two years presented at the MCH unit II PIMS



Figure-1: Patient's picture with new born baby.



Figure-2: B.P. cuff applied on lower limb & support given with wedge.

hospital, Islamabad, with achondroplastic features on May 15, 2018. She was a medical graduate and was a student of MPhil in psychology. Her age of menarche was 14 years and she had a regular menstrual cycle. There was no family history of achondroplasia as her parents and siblings were normal. None of her first degree relatives had achondroplasia.

Her husband was 32 years old and also a case of dwarfism (achondroplasia). She had no complications like respiratory or cardiac disorders.

She presented to our antenatal booking clinic at 31 weeks of gestation. Her LMP was 5/11/2017. There was no

significant past surgical history, with uneventful pregnancy. She was taking prenatal iron and vitamins supplements.

On examination, her weight was 48kg and height was 98cm; she appeared as a well-oriented normal intelligent woman with a large head and short limbs. She had slight kyphoscoliosis, enlarged tongue and protruded chin (Figure-1). She was mildly anaemic, and the thyroid gland was normal. On examination, the breasts were well-developed and revealed normal pregnancy changes.

Musculoskeletal system examination was normal with heavy bulk of muscles on the thighs. No abnormality was observed on abdominal examination and she had normal-looking external genitalia.

Her routine antenatal investigations, i.e. ECG and ECHO, were done for anaesthesia fitness and were found to be within normal limits.

Blood CP report showed Hb% 9.3 gm/dl, ultrasound scan done on June 23, 2018 revealed a single live foetus with parameters of 35 ± 2 weeks and no gross anomaly was seen.

For regular antenatal follow up, she visited the OPD at 34 weeks of gestation with complaints of discomfort in her regular activities and requested early delivery. She was counselled and a date was given for elective Lower Segment Caesarean Section at 37 weeks.

She was admitted at 36 ± 4 weeks and her anaesthesiology, neonatology and neurology consultations were done. She was very nervous and did not want to be awake during the procedure. Options for anaesthesia were discussed with the patient and she agreed for general anaesthesia. All the risks of general anaesthesia were explained in detail and written informed consent was taken. Facemasks, tracheal tubes, and laryngeal airways of appropriate size were prepared before the operation for possibility of difficult intubation. Preoperatively she was given antiemetic and anxiolytics.

Her Caesarian section was performed by the consultant on June 25, 2018 at 37 weeks of gestation under general anaesthesia by a senior anaesthetist and she was monitored throughout the procedure; due to her short limbs we applied B.P. cuff on her lower limb (Figure-2). The anaesthesia was given with Propofol and neuromuscular block was done with Succinylcholine. The patient was put in semi-flexed position, lower limb support was given with wedge during delivery the baby needed extra support and skills because of less space and flexed legs (Figure-2). A male baby, weighing 2.7 kg was delivered with Apgar score 8/10 and 9/10.

Intra-operative findings were: the uterus size, shape and lower uterine segment was well-developed and the

placenta was normal and of average size. No complications occurred during and after the Caesarian section. The baby was normal, externally examined by the paediatrician and was discharged.

Discussion

Achondroplasia is the commonest cause of dwarfism in which limbs are shorter than the trunk. The patients have typical achondroplastic facial features with a prominent forehead (frontal bossing) and a flattened bridge of the nose, enlarged skull growth (macrocephaly) and abnormal spinal curvature (kyphosis).⁷

Many comorbidities are associated with dwarfism; hence, it is necessary to assess the patient before anaesthesia, and caesarean section should be the preferred mode of delivery due to congenitally small and contracted pelvis.⁸

Limbs, craniofacial, skeletal abnormalities and spinal stenosis are associated with dwarfism. Problems may also occur in the central nervous system, respiratory and cardiovascular systems.

There are various methods for prenatal diagnosis of achondroplasia in the foetus, such as chorionic villus sampling at 10-13 weeks and amniocentesis at 15-18 weeks for analysis of DNA for FGFR3 gene mutation in foetal cells. It is also diagnosed by detailed ultrasound at 18-20 weeks if short limbs and facial features of achondroplasia are present.⁴

Selection of proper mode of anaesthesia, general or spinal, depending upon the patient's situation and condition is also important.^{6,8,9}

During airway management, the typical facial features of the achondroplastic patients may alert the anaesthetist regarding possible problems. Because of the large tongue and protruded jaw, the anaesthetist may have difficulty in maintaining upper air way patent. Endotracheal intubation may possibly be difficult because of shorter skull base with limited extension.^{6,7}

The anaesthetist may face difficulty during regional anaesthesia due to anatomical deformities in these patients, e.g. kyphoscoliosis, narrow vertebral canal, short pedicles, reduced interpeduncular distance and osteophyte formation, making spinal anaesthesia complicated; spinal stenosis may also impair the CSF flow. For epidural block, more difficulty can occur in identification of dural puncture due to narrow epidural space.⁶ Kyphoscoliosis also causes cardiopulmonary compromise in such patients. In addition physiological changes of pregnancy such as enlarged breast, airway oedema, increased plasma volume, increased oxygen

consumption and aortocaval compression by enlarged uterus can affect the health status of these patients.¹⁰

Spinal anaesthesia is very impredictive; decreasing intrathecal dose is not reliable and if given in high doses there is always a chance of high spinal.

Combined spinal and epidural technique may be a better choice with low dose spinal and titration of epidural anaesthesia, which causes rapid onset of spinal anaesthesia. This method can provide prolonged anaesthesia and good analgesia during post-operative period.^{6,11}

The height of our patient was 98 centimetres, without any co morbidity, and the new born was healthy, with 2.7 kg weight, no external abnormality was seen, and he was not admitted to the NICU. Both the mother and baby were discharged on the fourth day after the operation.

Conclusion

Since these patients are considered high risk, detailed prenatal counselling, comprehensive evaluation of the potential risks, and obstetric and perioperative management should be performed by a multidisciplinary care team, including an obstetrician, anaesthesiologist, pulmonologist, cardiologist and neonatologist. The mode of anaesthesia should be chosen according to the case and details of risks during anaesthesia must be clearly discussed with the patient with informed written consent taken from the patient or attendants. This case shows that it is possible for achondroplastic woman to become pregnant and deliver a healthy baby at term with proper management.

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