SUCCESSFUL TWO PREGNANCIES IN ACHONDROPLASIC MOTHER, A CASE REPORT

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INTRODUCTION

The word achondroplasia is derived from Greek and means "without cartilage formation," although individuals with achondroplasia do have cartilage.

In genetics, **dominance** describes the effects of the different versions of a particular gene on the phenotype of an organism. Many animals (including humans) and plants have two copies of each gene in their genome, one inherited from each parent.

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The different variants of a specific gene (such as that coding for earlobes) are known as alleles. If an organism inherits two alleles that are at odds with one another, and the phenotype of the organism is determined completely by one of the alleles, then that allele is said to be dominant. The other allele, which has no tangible effect on the organism's phenotype, is said to be **recessive**.

Achondroplasia is a genetic disorder of bone growth that is evident at birth and inherited as an autosomal dominant trait but most cases (80%) are due to mutations of fibroblast growth factor receptor 3 (FGFR3) (1, 2 , 3,4). These individuals have normal mental and sexual development, and life span may be normal**.** (5) It affects about 1 in 26,000 to 1 in 40,000 births (5, 6, 7), and it occurs in all races and in both sexes. Its depiction in ancient Egyptian art makes it one of the oldest recorded birth defects.

It is, however, the commonest cause of short-limbed dwarfism. It is a genetic disorder

Achondroplasia can be diagnosed on the basis of characteristic clinical and radiographic findings in most affected individuals. In infants, in whom the diagnosis can be difficult, and with individuals with atypical findings, molecular genetic testing can be used to detect a mutation in the *FGFR3* gene (locus 4p16.3). Such testing detects mutations in 99% of affected individuals and is available in clinical laboratories. (6, 7)

Certain gynaecological problems like infertility, menorrhagia, dysmenorrhoea, leiomyomata and early menopause are more common in these patients. Information regarding obstetric behaviour in achondroplastic females is scarce in literature. However, problems such as pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis necessitating lower section caesarean section, prematurity and foetal wastage, etc, have been reported. General anaesthesia is preferred to regional anaesthesia because of the spinal abnormalities.

There is increased neonatal mortality due to hydrocephalus and thoracic cage abnormality. Such a patient is considered high risk in terms of anaesthesia and obstetric outcome and there is enough room for prenatal counselling and diagnosis. (5).

Herewith we report a rare case of achondroplasia presented in Wad Madani Maternity Hospital, delivered towise by lower segment cesarean section, outcome was satisfactory to both mother and fetus.

CASE REPORT

A lady of 23 years old, received 8 years of basic education with an average performance, known case of a achondroplasia. Gravida two, Para one presented in labour to the outpatient clinic in Wad Medani Maternity Hospital.

Full history and complete medical examination reviewed. Her previous pregnancy ended with elective cesarean section. She was on regular antenatal care, and planned for elective cesarean section, which was carried out at 38 week of gestation. The operation was complicated with pulmonary edema and she found difficulties to recover from anesthesia, but in spite of that she had smooth postoperative period, and she was discharged home with her baby on 10^{th} postoperative day in good health. From that time she disappeared for one year, to present in labour. Uncertain of her dates but the clinical examination showed a fetus of 38 weeks and further the ultrasound scan confirmed the gestation age.

She was not known to be diabetic, hypertensive, asthmatic or have any significant medical problems. Her past medical and drug histories were not significant. But she had interesting family history. Her father is of 55 years old (Gaali tribe) ,known case of achondroplasia and known diabetic , has two wifes , and four sons (two of them are achondroplasic) , and six daughter (3 of them are achondroplasic) .His oldest daughter has a son and one daughter, all are normal . Our patient is the third member in family. She was married for three years, has two sons ,the older two years old and he is normal , the second one is 9 days and he is normal also.

The mother of our patient is the second wifes, no blood relationship with the father (Gwamaa tribe), 40 years old, not achnodroplasic and has no family history of achondroplasia and has no significant medical problems.

Clinical examination revealed that, 50 cm tall looked well, in pain not pale or jaundice, conscious and oriented. Had a massive skeletal deformities; chest and pelvis were deformed. Vital signs within normal ranges. Obstetrical examination showed a fundal height at 38 week, longitudinal lie, head not engaged, and

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fetal heart sound was detected .and there were marked abdominal contractions. Per vaginal examination confirmed that this lady was in labour .A decision of emergency cesarean section was taken, and proceeded toward team consultation .The team consisted of senior consultant obstetrician, endocrinologist and consultant anaesthetist .Decision of emergency cesarean section was taken. The operation was done under general anesthesia. We faced no difficulties to perform the operation, but we wer quite careful for the anesthesia and fluid overload. There were no preoperative, intraoperative or postoperative complications. Delivery was attended by paediatrician who received and examined the baby and commented that the baby was normal and achondroplasic.

The postoperative period passed smoothly, and she was discharged home with her baby on 10^{th} postoperative day in good health.

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DISCUSSION

Short-limbed dwarfism involves shortening of distal, middle or proximal part of the limbs (acromelic, mesomelic, and rhizomelic subtypes, respectively). Achondroplasia is the commonest form of rhizomelic dwarfism. This autosomal dominant disorder is associated with abnormal endochondral ossification whereas periosteal and intramembranous ossifications are normal (8).

These patients have peculiar facial features, bony deformities and systemic abnormalities that often make administration of anesthesia challenging. There are several uncertainties regarding the mode of anesthesia, exact procedures and protocols, drug choice and dosage, etc**.** (9-10) .There is only one published report from India**.** (11) Spinal anesthesia, on the other hand, has rarely been used, with only 4 published case reports till date. (10, 12, 13, 15) **,** This patient suffered from general anathesia in first CS ,and developed severe pulmonary oedema due to fluid overload , that was avoided in the second operation by fluid restriction which was adjusted according to body weight.

However, other than hypotension, no other serious adverse events, including he much-feared neurological deficits, have so far been reported.(14)

Regarding the outcome, the baby was quite normal with Apgar score 9 out of 10. The literature showed the approximately 7.5% of infants with achondroplasia die in the first year of life from obstructive apnea or central apnea. (16) Obstructive apnea may result from midface hypoplasia. Brainstem compression is common and may cause abnormalities of respiratory function, including central apnea. In one study, 10% of infants had craniocervical junction (CCJ) compression with abnormality of the cervical spinal cord. (17**)** All children who underwent surgical decompression of the CCJ had marked improvement of neurologic function.

Obesity is a major problem in patients with achondroplasia. Excessive weight gain is manifest in early childhood. Until a height of about 75 cm is reached, the mean weight-to-height ratios for children with average stature and for children with achondroplasia are virtually identical. Above a height of 75 cm, the weight-to-height ratio for patients with achondroplasia exceeds that of the general population**.** In adults, obesity may increase the morbidity associated with lumbar stenosis; in addition, it may contribute to non-specific joint problems and possibly to early mortality from cardiovascular complications**.** (16)

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