CASE REPORT

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Achondroplastic pregnancy A Case report

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DR. SHAHNAZ SADIQALI SYED[1]
DR. SYED SADIQALI ABASALI[2]
1. Clinician (Obstetrics & Gynecology), Miraj
2 Assistant Professor, GMC, Miraj

Corresponding Author:

Dr. Shahnaz Sadiqali Syed
Clinician (Obstetrics & Gynecology)
Miraj-416410 (Maharashtra, India)
+919421220419
shahnazsyed77@gmail.com

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Abstract:

Achondroplasia is a form of short-limbed dwarfism. It is characterized by short stature, affecting the growth of limb bones, spine and skull. It is inherited as autosomal dominant trait and most cases (80%) are due to mutations of fibroblast growth factor receptor 3 (FGFR3). Characteristic phenotypic features include short height, short upper arms and thighs, large head and forehead with a flat bridge of the nose, dental problems, broad and flat feet, short toes and short fingers, trident hand (a condition in which you there is an extra space between the middle and ring fingers), weak muscle tone. If there is pregnancy in achondroplastic female, there will be cephalopelvic disproportion and requires lower-segment caesarean section (LSCS). In present case a 23 year old achondroplastic dwarf given birth to normal male of 2500 g.

Keywords: achondroplasia, pregnancy, phenotype

Abbreviations: LSCS- lower segment caesarean section

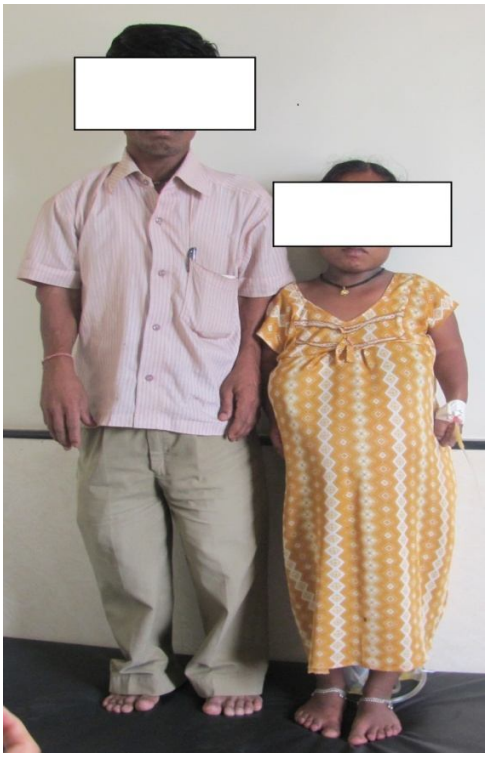
Introduction:

Achondroplasia, is the most common form of skeletal dysplasia characterized by short limb dwarfism, affecting the growth of limb bones, spine and skull. The prevalence is approximately 1 in 25,000.[1] It is inherited as autosomal dominant trait and most cases (80%) are due to mutations of fibroblast growth factor receptor 3 (FGFR3). People with achondroplasia can be born to parents that do not have the condition due to spontaneous mutation.[2] Characteristic phenotypic features include disproportionate short stature, megalencephaly, a prominent forehead (frontal bossing), midface hypoplasia, rhizomelic shortening of the arms and legs, a normal trunk length, prominent lumbar lordosis, genu varum, and a trident hand configuration.[3]

Case report:

Achondroplastic dwarf patient registered to outpatient department in Mamta Nursing Home, Miraj with history of 2 months of amenorrhea. Her urine pregnancy test was positive. Ultrasonography was done for gestational age and viability. It suggested of 8 week pregnancy.

The patient married to achondroplastic male 1 year back. The marriage was non consanguineous. On examination patients general condition was average. Patient height was 104 cm and weight was 25 Kg. Her intelligence was normal. Physical examination shows large head and short limb. Blood pressure, pulse rate and temperature were in normal range.

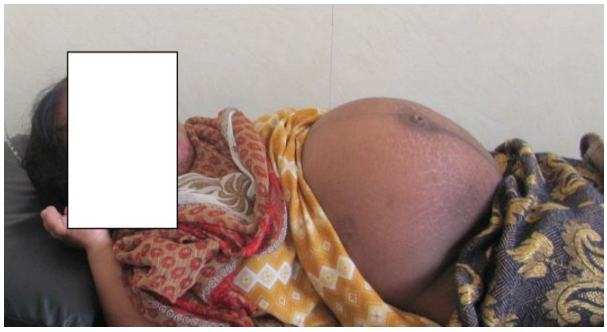


Photograph 1. Patient with her husband

On doing per abdominal examination, uterus was palpable, even though it was only 8 week pregnancy, normally which is palpable after 12 weeks of pregnancy. All investigations were done and were normal. All risk associated with pregnancy and also obstetric and anaesthesia problems in achondroplasia were explained to her, husband and relatives.

Patient was having regular antenatal checkups. At 20 weeks anomaly scan was done which was normal and showing normal growth pattern. Her total antenatal period was uneventful. Fetal growth was absolutely normal. At term lie of baby was oblique with vertex in left iliac fossa. An elective lower-segment caesarean Section (LSCS) was planed at 37 weeks.

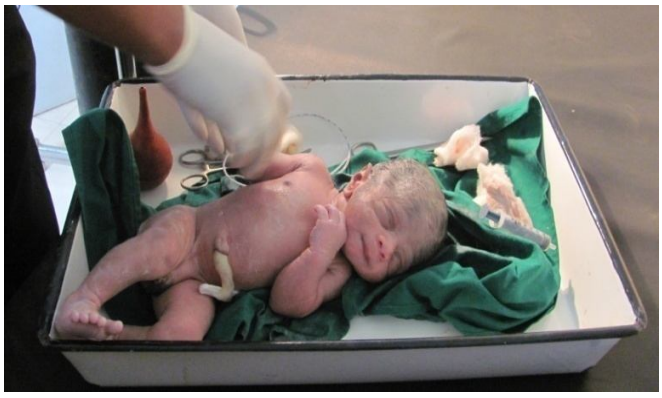
Before LSCS, opinion of physician, E.N.T Surgeon and Anesthetist were taken. Her spirometry study shows restrictive pattern and ECG shows sinus tachycardia. Even though her spine was having Kyphoscoliosis, spinal anesthesia was planed.



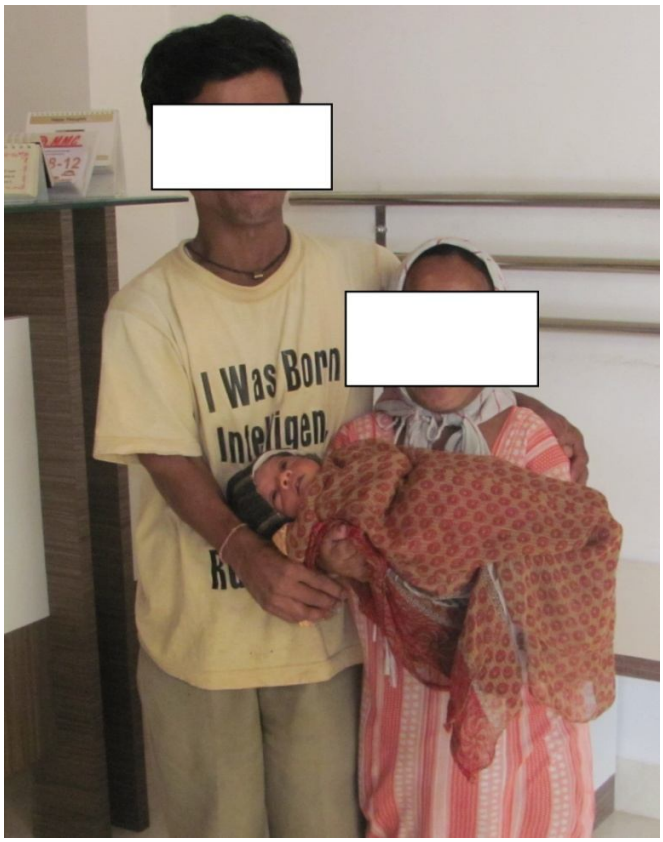
Photograph 2. Patient at full term

Photograph 3. LSCS procedure

At operation, spinal anesthesia was given with lignocane, Heavy 5%. It was uneventful. LSCS was performed by transverse Pfannenstiel incision incision. Procedure was uneventful and a live male baby weighing 2,500g was delivered. Pediatrician examined the neonate and ruled out achondroplasia in baby.



Photograph 4. Neonate examined by pediatrician



Photograph 5. Patient at the time of discharge

Discussion:

Achondroplasia is a bone growth disorder that causes disproportionate dwarfism. Achondroplastic female have pelvic diameter smaller than normal. If there is pregnancy in Achondroplastic female, there will be cephalopelvic disproportion as size of fetal head will be normal. So normal delivery in these patients is not possible and requires LSCS. In present case also LSCS was performed by transverse Pfannenstiel incision and entire procedure was uneventful.

Achondroplastic patients have a number of anatomical and physiological abnormalities that contribute to problems with the administration of obstetric anesthesia and performance of caesarean section. In present case spinal anesthesia was given, even though her spine was having kyphoscoliosis. There were no problems with the administration of anesthesia.

If both parents are affected with Achondroplasia then 25% children will be normal, 50% heterozygous and 25% will be homozygous mutation. Homozygous achondroplasia is always lethal. In present case pediatrician examined the neonate and ruled out achondroplasia in baby.

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