OLGU SUNUMU CASE REPORT

Maternal and Fetal Outcome of a Pregnancy in an Achondroplastic Dwarf: Case Report

Akondroplazik Gebeliğin Maternal ve Perinatal Sonuçları

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ABSTRACT Achondroplasia is a genetic disorder of bone growth that is evident at birth. It affects about one in every 25 000 births and it occurs in all races and in both sexes. These cases have normal mental and sexual development, and life span may be normal. Problems such as pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis necessitating caesarean section, prematurity, and fetal wastage are more common in achondroplastic pregnant females. An achondroplastic pregnant case who has the shortest height and the oldest age in the related literature was presented. The important points on management of achondroplastic pregnancies and their complications were discussed.

Key Words: Achondroplasia, pregnancy

ÖZET Akondroplazi doğumda ortaya çıkan genetik bir kemik büyüme bozukluğudur. Tüm ırklar ve her iki cinste olur ve yaklaşık 25 000 doğumda bir görülür. Bu olgular normal mental ve seksüel gelişme gösterir ve normal bir yaşam beklenir. Akondroplazik gebe kadınlarda preeklampsi, polihidramniyoz, solunum sıkıntısı, sezaryen gerektiren dar pelvis, prematürite ve fetal kayıp daha sık görülür. Literatürdeki en yaşlı ve en kısa boya sahip bir akondroplazik gebelik olgusu sunuldu. Akondroplazik gebeliklerin ve komplikasyonlarının yönetimindeki önemli noktalar tartışıldı.

Anahtar Kelimeler: Akondroplazi, gebelik

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Achondroplasia is the most common form of inherited disproportionate short stature. It occurs in one in 15.000 to 40.000 live births.[1] The worldwide population of dwarfism is about 190.000.[1] Approximately 150.000 persons have achondroplasia in the entire world.[1] Achondroplasia is characterized by abnormal bone growth that results in short stature with disproportionately short arms and legs, a large head, and characteristic facial features with frontal bossing and mid-face hypoplasia. Average adult height for men with achondroplasia is 131±5.6 cm; for women, 124±5.9 cm.[1] Most individuals with achondroplasia are expected to have normal mental and sexual development and a normal life expectancy. Diagnosis is made by physical examination and skeletal radiographic findings. Achondroplastic women are able to conceive. However, achondroplastic pregnant women are considered at high risk.

Here a case of achondroplasia with pregnancy was reported. The patient, an achondroplastic dwarf had a caesarean section within 33 weeks of pregnancy. This case was the shortest and the oldest achondroplastic pregnant in the literature.

CASE

A 41-year-old, gravida 2 para 0 , achondroplastic pregnant admitted to our Gynecology and Obstetrics Clinic at 25 weeks of gestation due to dyspnea . She had not any antenatal care. She has been married for 3 years, has regular menstrual cycle, and didn't use any contraception. The couple was nonconsanguineous. There was not any special feature on her gynecological history except smoking. Her mother was at normal stature, but her father was achondroplastic. Her husband was 37-year-old, 187 cm tall, healthy and without family history of skeletal dysplasia.

Physical examinations revealed a 37 kg weighting, 100 centimeters tall, healthy female with typical features of achondroplasia including large head, short limbs and marked kyphoscoliosis of thoracolumbar spine (Figures 1, 2). Blood pressure, heart rate, and body temperature were normal. The cardiovascular system examination revealed first degree failure of mitral valve. She also suffered from dyspnea because of bronchitis.

FIGURE 1: The side-view of achondroplastic pregnant in 32nd gestational weeks.

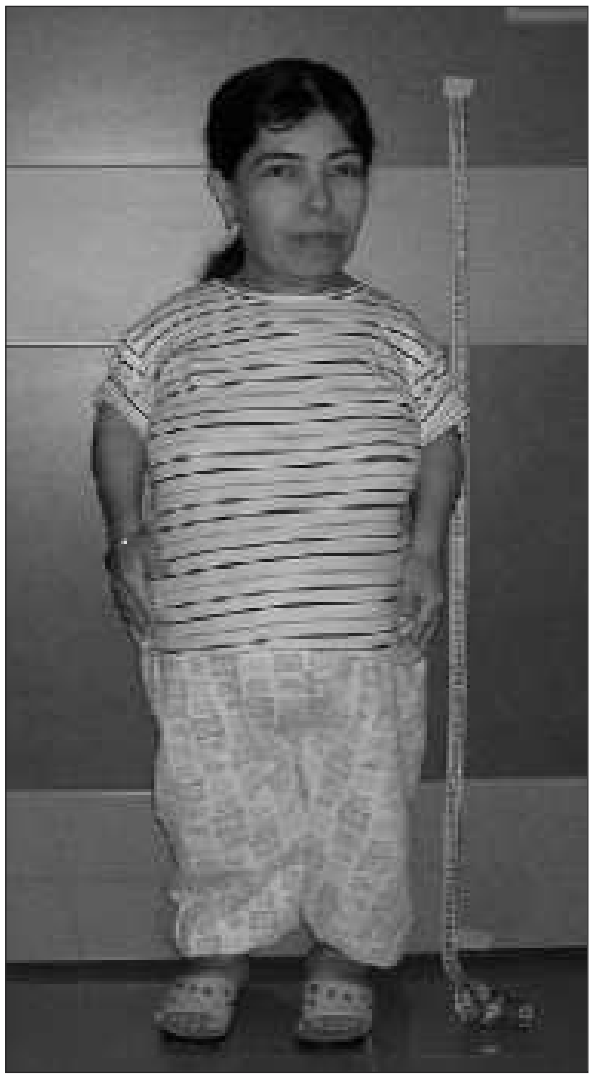

FIGURE 2: The front-view of achondroplastic pregnant in 32nd gestational weeks.

Medical treatment and oxygen supplement was administered for bronchitis. Her dyspnea was cured after treatment and her pregnancy was monitored in the hospital until delivery. The fetal ultrasonic findings were normal. Umbilical and uterine arteries Doppler parameters were normal. For fetal lung maturation, bethamethasone 12 mg/24 h (Celestone chronodose®, Schering Plough, Kirklareli, Turkey) was administered in 28th gestational weeks and repeated in 32nd gestational weeks.

The premature rupture of membranes was occurred at the 33 weeks of her pregnancy. She had contracted pelvis because of achondroplasia, so a caesarean section was performed as soon as possible. The operation was performed under general anesthesia. A healthy male infant weighing 1680 g, measuring 49 centimeters with an Apgar score of 7 at 1 min and 9 at 5 min was delivered. After the pediatric examination of the infant, no feature of achondroplasia was found. She had a normal postpartum course. Mother and baby were discharged from hospital on day 7 without any complication.

Written and informed consent was taken from the patient for publication.

DISCUSSION

Achondroplasia is a common, nonlethal form of chondrodysplasia. It is a genetic disorder and inherited as an autosomal dominant trait but most of cases (75-80 %) are due to de novo mutations. Achondroplasia is caused by a mutation in the gene for fibroblast growth factor receptor-3. The gene has been mapped to band 4p16.3. A lot of researches are being done in this area. [2,3] In our case, the disease was inherited as autosomal dominant trait, because her father was achondroplastic.

An individual with achondroplasia who has a reproductive partner with normal stature has a 50% risk in each pregnancy of having a child with achondroplasia. If the partner also has short stature, the specific recurrence risk and the possibility of a severely affected infant must be determined.[4] Prenatal diagnosis for high risk pregnancies is possible by analysis of DNA extracted from fetal cells obtained by amniocentesis usually performed at about 15-18 weeks' gestation or chorionic villus sampling at about 10-12 weeks' gestation.[5,6] The disease-causing allele(s) in the affected parent or parents must be identified before prenatal testing. The optimal time for determination of genetic risk and discussion of the availability of prenatal testing is before pregnancy. Our case admitted to our clinic at 25 weeks of gestation without having any genetic counseling, prenatal care and prenatal screening.

Achondroplastic dwarfs have normal sexual development, and low fertility rates. Certain gynecological problems like menorrhagia, dysmenorrhea, leiomyoma, and early menopause are more common in these patients. Contraception is important in achondroplastic women. Women with short stature using the oral contraceptive pill may have difficulty achieving the correct dosage for body weight even with current low dose preparations. Intrauterine device can exacerbate cramps and heavy flow in a group of women who already have a high incidence of these problems. Using barrier methods is difficult because of the short arms in achondroplasia. Allanson et al in a study with 150 cases found that the most common method used was rhythm method.[4] Finding the appropriate long-term contraception may require consultation with a gynecologist.

Previous studies have shown a higher proportion of women with achondroplasia to be childless and have reduced family size in comparison with average population.[7] They have a higher mean maternal age at conception. This may reflect delayed socialization and marriage with the partner. In our case advanced maternal age, delayed marriage age, and subfertility is concordant with the literature.

Information regarding obstetric management in achondroplastic females is insufficient in literature. Obstetric problems such as pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis necessitating caesarean section, prematurity and fetal wastage have been reported more in achondroplastic pregnants.[4,8] She had a spontaneous abortion in her history. Pre-eclampsia and polyhydramnios were not established at clinical and ultrasonographic findings.

In achondroplastic pregnancies, respiratory disorders are seen frequently, including apnea and restrictive pulmonary disease. Respiratory compromise especially is common during the third trimester. Baseline pulmonary function studies should be carried out before pregnancy to aid in evaluation and management.[4,9] Our patient had a respiratory problem when she was at the 25th weeks of pregnancy. A baseline pulmonary function study should also be done for the mother may develop respiratory compromise in the third trimester of pregnancy.

Because of limitation of uterine expansion, low birth weight found in infants born to women with short stature.[10] The infants with normal stature of women with achondroplasia are larger than those born to women with osteogenesis imperfecta and cartilage hair hypoplasia.[4] The infant was with normal stature and big for gestational age with birth weight in the 95th percentile, and birth height in the 95th percentile.

Because of small size of the pelvis in affected women, the preferred method of delivery is cesarean section without trail of labor. General anesthesia is preferred to regional anesthesia as the mother may develop complications due to spine stenosis. In our case, cesarean section was carried out because of preterm premature rupture of membranes and narrow pelvic outlet. Operation of our case was applied under general anesthesia without any complication.

In achondroplastic women; contraception and prenatal genetic counseling is also very important. These women are at high risk of unexpected maternal, perinatal and genetic complications, so their pregnancy should be managed carefully. In this paper we presented and discussed a case of achondroplastic pregnancy.

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