Successful Delivery in a Woman With Achondroplasia: A Case Report

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Abstract- Achondroplasia is an autosomal dominant disease which is characterized by limb shortening and narrow trunk, and macrocephaly. Women with achondroplasia suffer from infertility, menorrhagia, dysmenorrhoea, leiomyomata and early menopause. Our case was a 26-year-old woman with achondroplasia who had scoliosis and osteoporosis. She referred to our clinic at 9 weeks of gestation and had all screenings completely. She had a single female fetus with no abnormalities. She had an emergent due to rupture of membranes at 37 weeks and 3 days under general anesthesia. The neonate had no complications. The first minute Apgar score was 9 and 5th-minute Apgar score were 10. Umbilical artery Ph was 7.26 and birth weight was 3140. A woman with achondroplasia could have a normal pregnancy and give birth to a healthy term neonate under precise screening.

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Introduction

Achondroplasia is an autosomal dominant disease which has prevalence as 1:5000 to 1:40000 live births (1). Fibroblast growth factor receptor alteration is responsible for disease occurrence resulting cartilage growth change and endochondral ossification (1). Patients suffer from disproportionate dwarfism along with craniofacial, central nervous system, spinal, respiratory and cardiac anomalies (2). Infertility, menorrhagia, dysmenorrhoea, leiomyomata and early menopause are common in these cases (3).

Shorter proximal limb bones, narrow trunk, and macrocephaly are characteristics of patients with achondroplasia (4-6). Adults with achondroplasia have a fixed, angular thoracolumbar junction kyphosis which will lead to the respiratory and cardiac dysfunction that make difficulty in general anesthesia (2). So, airway management will be risky while general anesthesia is preferred to regional anesthesia due to spinal abnormalities. Here we report a case that had achondroplasia and had a successful delivery.

Case Report

A 26-year-old primiparous achondroplastic woman with height 98 cm referred to the prenatal clinic of Mohebe Yas hospital (affiliated Hospital of Tehran University of Medical Sciences). She had high school graduation and was working in the laboratory. Her menarches started at 16 years of age, and she had regular menstrual cycles.

She had right and left retinal detachment 10 years ago which led to vision loss of one eye completely and made the vision of the other eye weak. Scoliosis was present while she had no respiratory or cardiac anomalies. Severe osteoporosis was evident. Her husband was a 26-year-old healthy man.

When she referred to our clinic her gestational age was 9 weeks, and she attended all screening programs and followed up to delivery.

Her last sonographic examination on 2015.10 revealed a live single female.

Transverse fetus with FHR=133, FL=68, posterior placenta grade II. The amniotic fluid level was normal, and there was no abnormality.

She had an emergent caesarean section due to rupture of membranes at 37 weeks and 3 days under general anesthesia.

Before section, she had cardiac and respiratory consultation and airway examination. Ketamine 1 mg/kg, plus propofol 2.5 mg/kg and 2.5 mg succinylcholine chloride administered for anesthesia. Tracheal intubation was achieved with a 6.5 mm tracheal tube using a stylet.

She and her daughter had no complications and

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discharged. The first minute Apgar score was 9 and 5th minute Apgar score was 10. Umbilical artery Ph was 7.26 and birth weight was 3140.

Discussion

Achondroplasia is an autosomal dominant inherited disease which is the most common type of dwarfism (7). Due to comorbidities which are associated with dwarfism, anesthetic assessment is essential in such cases, and preferred mode of delivery is caesarean section according to congenitally small and contracted Pelvic (8,9). Cases with achondroplasia have short stature and shortened

Limbs, craniofacial, spinal, skeletal abnormalities with problems in the central nervous system, respiratory and cardiac systems (2).

The height of our case was 98 cm, and she had scoliosis and severe osteoporosis but had no other comorbidities.

To choose general anesthesia or spinal anesthesia, patient's situation and condition should be evaluated carefully, and proper mode of anesthesia should be selected.

As the expansion of thorax, circumference is limited during pregnancy in achondroplastic cases, and enlarged uterus of pregnancy will impact small thorax cage. On the other hand, kyphoscoliosis will reduce lung capacity more. All of these complications will result in respiratory distress during the third trimester and the need for early delivery (2).

Our case delivered at 37 weeks of gestation while in Dubiel *et al*., case report delivery occurred at 32 weeks of gestation. Spinal anesthesia due to kyphoscoliosis in these patients will be difficult and associated with neurological complications while general anesthesia with straightforward airway management will be safe with no damage to the cervical spine.

Intubation in this patient done easily as literature shows that due to large head and tongue, difficulties in extending the neck and cervical instability, hard intubation should be expected in achondroplasia cases (2). On the other hand, pregnancy changes of airway should be considered in these cases.

The newborn of our case was healthy; she had no low birth weight, no icter, and had not admitted to NICU.

A woman with achondroplasia could have a normal pregnancy and give birth to a healthy term neonate under precise screening.

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