Case Report

Pregnancy in achondroplastic woman

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Achondroplasia is an autosomal dominant chondrodysplasia. The defect is in the process of conversion of cartilage to bone. People with achondroplasia seldom reach 5 feet in height. Intelligence is in the normal range. During pregnancy there are lot obstetric complications. There is no specific treatment for achondroplasia. Here is a case of achondroplastic woman delivering achondroplastic newborn depicting both autosomal and spontaneous mode of inheritance.

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Key words **: Pregnancy in achondroplastic woman, autosomal dominance.**

Achondroplasia, a type of osteochondrodysplasia is the common cause of disproportionate dwarfism. The prevalence is approximately 1 in 15,000 to 40,000. Achondroplasia may be inherited as an autosomal dominant trait or spontaneous mutation. 75% of achondroplastic babies are born to normal parents.

Case Report

A 30 years old unmarried, unbooked, primigravida from low socio-economic status, reported to our hospital with history of 8 months amenorrhoea. LMP was not known. She is 10th born to normal statured parents. Other siblings are normal statured. She is a known epileptic on carbamazepine.

Examination **—** On examination she is a dwarf. Height measures 113cms . Weight is 38kgs. BMI:29.8. She is anaemic. She has prominent forehead with frontal bossing, depressed nasal bridge and proximal limb shortening. Gravid uterus with fundal height corresponds to 32 weeks. Fetus is in cephalic presentation. Fetal heart sounds present. Fluid thrill present (Figs 1&2).

Fig 1 — Achondroplastic mother compared with normal mother

Fig 2 — Relatively large skull with depressed nasal bridge

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Antenatal ultrasonogram (Figs 3-6) :

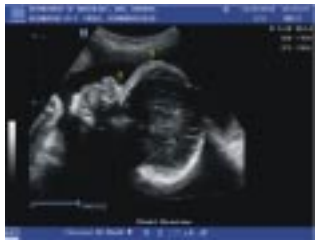


Fig 3 — Relatively large skull with narrow skull base and depressed nasal bridge

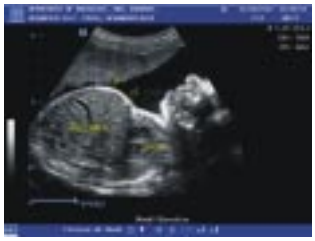


Fig 4 — Comparatively small thorax with protuberant abdomen

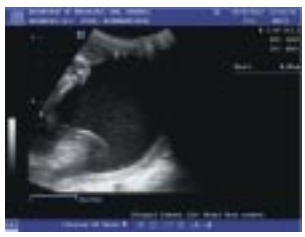
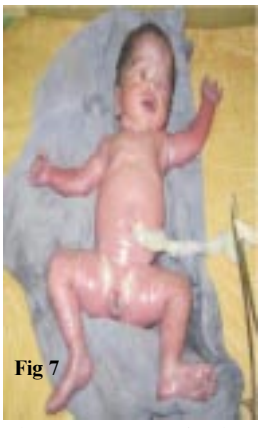


Fig 5 — Polyhydramnios

Fig 6 — Fetal biometry showing small long bones

Examination of Fetus (Figs 7&8) :



Figs 7 & 8 — Fetus showing relatively large skull, short limbs, depressed nasal bridge, small thorax and protuberant abdomen

A detailed antenatal ultrasonogram was done to rule out skeletal dysplasia. It revealed a live intra uterine fetus of gestational age around 30 weeks. The fetus had relatively large head with narrow skull base, frontal bossing, depressed nasal bridge, small chest with protuberant abdomen, short long bones (24 weeks) and normal kidneys which confirmed non-lethal skeletal dysplasia. Amniotic fluid was increased in quantity and AFI was around 22.

Emergency CS done under GA at the onset of labour pains, delivered a live preterm female baby with skeletal dysplasia confirming the USG report. Birth weight 1.5 kgs Baby was under the care of neonatologist & died after 6 hours of birth.

Discussion

Achondroplasia is caused by mutation of fibroblast growth factor receptor 3 (FGFR3) on chromosome 4[1]. New gene mutations leading to achondroplasia are associated with increasing paternal age as in our case. These individuals have normal mental and sexual development, with normal life span. Obstetric problems are foetal wastage, prematurity, pre-eclampsia, polyhydramnios[2], respiratory compromise, contracted pelvis necessitating lower segment caesarean section.

Gynaecological problems like infertility, menorrhagia, dysmenorrhoea, leiomyoma and early menopause are more common in these patients. General anaesthesia is preferred to regional anaesthesia because of the spinal abnormalities[3]. Gene based therapy is on the horizon.

Conclusion

Achondroplsia is one of skeletal dysplasia with normal IQ. Homozygous cases are lethal. In 75% cases occurs as spontaneous mutation . Antenatal USG is important. Such a patient is considered high risk in terms of anaesthesia and obstetric outcome and there is enough room for prenatal counselling and diagnosis. Incidence of unwed pregnancies are increasing nowadays. Women of low socio economic groups & disabled are exploited more. Women must be safe guarded from these exploitations.

References

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