THE ANTENATAL AND INTRAPARTUM MANAGEMENT OF WOMEN WITH ACHONDROPLASIA

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Achondroplasia is an autosomal dominant condition associated with short-limbed dwarfism. It is caused by a mutation in the fibroblast growth factor receptor-3 gene, with 80% being de novo. It affects roughly 1 in 22,000 babies born worldwide. There is limited information regarding the management of pregnant women with achondroplasia.

CLINICAL FEATURES

- Large head and frontal bossing
- Small mid-face and flattened nasal bridge
- Short limbs and spinal deformities

ANTENATAL CARE

Genetic counselling is important to discuss the risk of a baby inheriting achondroplasia.

One parent with achondroplasia	*Both parents with achondroplasia*
50% risk of an affected child	50% risk of an affected child 25% risk of homozygous achondroplasia (lethal)

Pre-natal testing should be offered

Serial growth scans are indicated as ultrasound features of achondroplasia are only identified later in pregnancy.

Anaesthetic review allows women to be counselled about difficult regional analgesia due to spinal deformities, and risk of general anaesthesia.

Baseline lung and cardiac function should be tested to identify any abnormalities.

Occupational therapy and Physiotherapy assessment help in the management of pelvic pain and postnatal adaptations.

INTRAPARTUM CARE / DELIVERY

The majority of women with achondroplasia deliver by caesarean section due to a narrow pelvis and risk of cephalopelvic disproportion.

Difficulty delivering the head in the context of fetal achondroplasia should be anticipated

⩽39

Delivery is commonly performed before 39 weeks due to spontaneous labour or increasing maternal discomfort.

A neonatologist should be present at delivery due to the increased risk of respiratory compromise and difficult resuscitation if the baby has achondroplasia.

SUMMARY

Pregnancy in women with achondroplasia is high risk and requires a number of antenatal considerations such as pre-natal testing, serial growth scans and baseline physiology testing. A multidisciplinary approach involving obstetricians, anaesthetists, geneticists, occupational therapists, physiotherapists, and neonatologists is key.