

Antenatal Diagnosis of Grebe Syndrome in a Twin Pregnancy by Ultrasound

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Grebe syndrome is a nonlethal distinct type of dwarfism in which a relatively normal head and trunk contrasts strikingly with a phocomelic malformation of the four extremities; the severity of the anomalies progresses distally along the limbs, so that the fingers and toes loose their phalangeal appearance and become mere knobs(I). Polydactyly is present in about 60% of cases. This disorder is inherited as an autosomal recessive trait(2). Although a number of cases have been described postnatally, we do not know of any case diagnosed antenatally. In our previous report of a family with Grebe syndrome, we had mentioned about the possibility of prenatal diagnosis of Grebe syndrome by ultrasound(3).

Prenatal detection of many skeletal dysplasias has been reported earlier as in anchondroplasia, spondylo thoracic dysplasia, diastrophic dwarfism, short rib-polydactyly syndrome type II Mejewski, Ellis-Van Creveld syndrome and many others(4).

We report probably the first case in the world, of an antenatal diagnosis of Grebe syndrome in one of the twins, the mother of whom had two earlier children with Grebe syndrome.

Case Report

A 32 years old gravida-4, para-3, was subjected for ultrasound examination at 26 weeks of gestation to rule out the possibility of Grebe syndrome, as she had two earlier children affected with Grebe syndrome. The recurrence risk in the present pregnancy was 25%, as Grebe
syndrome is an autosomal recessive disorder. A diamniotic dichorionic twin pregnancy was diagnosed. One of the fetuses was female and had BPD of 72 mm and femoral length of 54 mm, which corresponded to 26 weeks of gestation and the other male fetus showed shortening of all the 4 limbs (Figs. 1 a, b & c) with marked involvement of lower limbs and absence of fibula with a normal head and body contour. The femoral length was 23 mm (<5th centile), tibia 22 mm (<5th centile), humerus 39 mm (<5th centile) and BPD was 74 mm (50th

Fig. 1a. Ultrasound film showing marked shortening of femur.

Fig. 1b. Ultrasound film showing short tibia with absent fibula.
centile)(5,6). Fingers and toes were severely affected and appeared like knobs. The pregnancy was continued and she delivered twins at term. Both the babies were subjected to clinical and radiological evaluation. The male baby weighed 3.1 kg and had deformities of the limbs. Upper limbs were short, forearms being shorter than the upper arms and his fingers were bulbous. Lower limbs were shorter than upper limbs, the severity of shortening increasing distally. The feet were short and broad with valgus deformity. The toes were short and bulbous (Figs. 2a & b). X-ray showed thickening and shortening of long bones, arrested

Fig. 1c. Ultrasound film showing shortening of arm and forearm bones.

Fig. 2a. Twin on the right with Grebe syndrome.
growth of ulna, single phalanx in most of the digits, absence of fibula and malposition of tarsal bones (Fig. 3). The female baby weighed 2.7 kg and showed no clinical or radiological evidence of dysplasia.

Discussion

Prenatal diagnosis of bone dysplasia presents difficult challenges for clinician involved in monitoring pregnancies. The growing use of ultrasonogram for fetal monitoring has increased the number of skeletal dysplasias diagnosed antenatally. Even though prenatal diagnosis of bony anomalies are possible, it poses a great difficulty in differentiating one skeletal disorder from the other, that an overt clinical inspection can offer, which makes bone dysplasia one of the most challenging diagnosis in the prenatal ultrasonography(4).

In our case we were able to diagnose
Grebe syndrome antenatally because of history of previous two siblings affected with the same disorder and by comparing BPD and length of limb bones. In the affected male fetus the BPD was on the 50th percentage, whereas all the other long bones measured were less than 5th percentile (5,6), thus indicating limb reduction anomaly in the fetus. The family history of 2 previously affected sibs was an important pointer for the diagnosis. This only emphasizes the importance of good family history and study of members of family before attempting prenatal diagnosis by ultrasonography.

Ultrasonological features noted in our case included shortening of all 4 limbs, lower limb bones shorter than upper limb bones, the severity of shortening increasing distally with short bulbous fingers. The high incidence of premature deaths accounting for 11% of stillbirths and 38% of infant mortality(7) and the extreme disability it produces in an affected individual, may warrant early detection by ultrasound and possibly termination of pregnancy.

REFERENCES


Lethal Forms of Short Limb Dwarfism

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Short limb dwarfism is part of tubular bone chondrodysplasia syndromes. It can manifest both in lethal and non-lethal forms(1). Short rib-polydactyly

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