Schneckenbecken dysplasia, radiology, and histology

Abstract To our knowledge this is the first report of Schneckenbecken dysplasia with the development of hydrops early in the second trimester. The radiological findings showed the typical hypoplastic iliac bones with medial extension and very flattened, on lateral view, oval-shaped vertebral bodies and short long bones. The histology showed hypercellular and hypertrophic cartilage with chondrocytes with centrally located nucleus. The absence of the lacunar space as described before was also observed in some chondrocytes in our case. This male fetus was the product of consanguineous parents of Mediterranean origin compatible with autosomal recessive inheritance.

Introduction
Several different types of lethal short-limbed skeletal dysplasia with platyspondyly have been recognized with a different mode of inheritance. Schneckenbecken dysplasia, a rare skeletal dysplasia, is one of them, with an autosomal recessive mode of inheritance [1]. Some of the lethal skeletal dysplasias have been reported to present with hydrops, e.g. achondrogenesis I-A, the Langer-Saldino dysplasia (achondrogenesis type II), the platyspondylic lethal chondrodysplasia Torrance and San Diego type, and some types of the short-rib (polydactyly) syndromes [2]. We describe a case of Schneckenbecken dysplasia that presented with hydrops and short-limbed skeletal dysplasia early in the second trimester.

Case report
We describe the clinical, radiographic, and histological features of a case of Schneckenbecken dysplasia. This was the second child of consanguineous parents of Mediterranean origin. On ultrasound at 20 weeks of gestation, a fetus was seen with severe hydrops, extremely short extremities, and a small thorax. Termination of pregnancy was performed at 22 2/7 weeks. The first child had a congenital fiber-type disproportion, an autosomal recessive disor-
**Fig. 1** Frontal and lateral view of the fetus. The severe hydrops and extreme shortness of the extremities are obvious.

**Fig. 2** Anteroposterior (A) and lateral radiograph (B). Note the medial extension on the right side of the ileum (C) (snail-like pelvis, Schneckenbecken), oval shape of the vertebral bodies on lateral X-ray and the dumbbell-like appearance of the short long bones.

der of skeletal muscle. At autopsy, a male fetus was seen with hydrops (Fig. 1), general shortness of the extremities, a central medial palatoschisis of the hard and soft palate and lung hypoplasia. The abdomen was very prominent. Body weight was 360 g, crown-rump length was 16 cm, and the crown-heel length was 18 cm. The post-mortem radiographs (Fig. 2) showed some enlargement of the biparietal diameter. The long bones were extremely shortened with a dumbbell-like appearance without apparent metaphyseal abnormalities and a short diaphysis. The vertebral bodies were flattened, hypoplastic, and on the lateral projection somewhat oval. There was marked lordosis in the lumbosacral region. The ribs were short. The scapulae were not abnormal. The medial part of the iliac wing on the left side showed a peculiar projection (snail-like pelvis/Schneckenbecken dysplasia; Fig. 2C). There was