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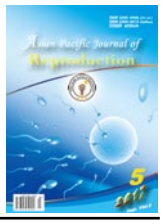

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A case report of prenatally detected achondrogenesis type Ⅱ with an occipital cephalocele

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ABSTRACT

Achondrogenesis is a very rare lethal skeletal disorder. Here we describe a case of prenatally diagnosed achondrogenesis type Ⅱ in a 28 year-old woman at (17+4) wk. She had history of 5 first trimester missed abortions. The couple is consanguineous. Ultrasonography showed extreme micromelia, short neck and trunk, large head and prominent abdomen. Delayed ossification in sacral bones was detected and ossification of pubic rami was poor. There were associated large cystic hygroma, anasarca and also high occipital cephalocele. Posterior fossa was normal. Mild hypothelorism, depressed nasal bridge, low set ear and mild retrognathia were identified too. Amniocentesis result was compatible with a normal female fetus. Post mortem whole body radiography confirmed the diagnosis. To our knowledge, this is the second case report with association of cephalocele and achondrogenesis type Ⅱ.

1. Introduction

Achondrogenesis is one of lethal type of skeletal dysplasia, which is the most severe form with extreme micromelia[1]. It is characterized by short trunk, protuberant abdomen, disproportionatly large cranium and some degree of endochondral ossification deficiency[2]. Hydrops fetalis and some facial features as retrognathia also have been reported as associated abnormalities. They are divided into two groups, type Ⅰ (A and B) and type Ⅱ mainly based on radiological and clinical studies such as rib fractures and pattern of poorly ossified bones[3]. Type Ⅱ is also called Langer-Saldino Achondrogenesis.

We report a case of type Ⅱ achondrogenesis with occipital cephalocele in an 18 wk fetus diagnosed on prenatal sonography and post mortem radiography.

2. Case report

A 28 years old gravida 6 para 0 at (17+4) wk was referred to our hospital with diagnosis of hydrops fetalis and tetra phocomelia. She

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had history of 5 first trimester missed abortions around 6-8 wk. The couple is first degree relative consanguineous without family history of skeletal abnormalities, teratogen exposure or any other disease.

Prenatal sonography at out center revealed, extreme micromelia (femur length: 4.4 mm, humerus length: 5 mm, tibia: 3.3 mm and ulna: 4.3 mm). Foot length (19 mm) was normal for fetal age and Femur length (FL)/foot ratio (0.23) was severely decreased. FL/abdominal circumference ratio (0.02) was also severely decreased, in favor of lethal skeletal dysplasia. There was evidence of associated bell shaped and narrow thorax, short neck and trunk, prominent abdomen [enlarged abdominal circumference: 156 mm, (20+6) wk] and macrocephaly [head circumference: 162 mm, (19+1) wk]. Delayed ossification in sacral bones was detected and ossification of pubic rami were poor (Figure 1A and B). The ossification of skull and ribs appeared normal and there was no evidence of rib or other bone fracture. There was evidence of cystic hygroma and hydrops fetalis (extensive skin edema and mild plural effusion). In the evaluation of head and neck, a high occipital meningocele, mild hypothelorism, depressed nasal bridge, low set ear and mild retrognathia were evident (Figure 1C).

This study was approved by the local committee for ethics in research and permission to carry out the study was obtained from the parents and the informed consent was taken from them. Amniocentesis was compatible with normal 46 xx karyotype. Based on the mentioned findings, achondrogenesis type Ⅱ was suggested. Parents chose termination of pregnancy and post mortem photography and babygram confirmed the diagnosis (Figure 2). The size of fetus in Figure 2 is 105 mm. In babygram extremely short limbs, narrow thorax with short ribs, short trunk, delayed ossification in sacrum, poor ossification of pubic bones and macrocephaly were evident. There was no bony fracture.

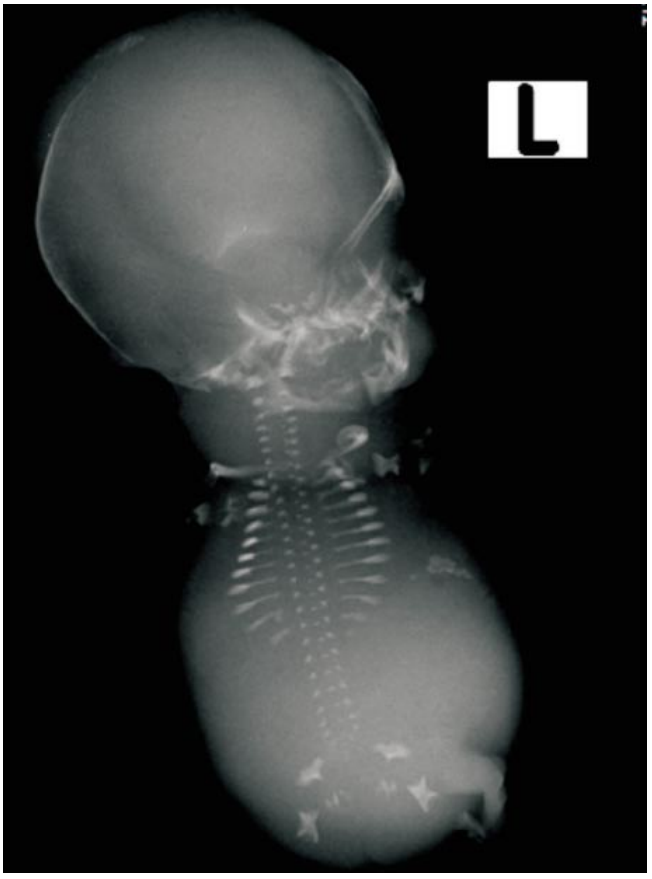

Figure 2. Post mortem baby gram showing delayed ossification of sacral bones, short trunk, macrocephaly and extremely short long bones.

3. Discussion

Achondrogenesis is an osteochondysplasia that affects the growing cartilages and is caused by mutations in collagen genes.

It includes that a heterogeneous group of skeletal dysplasia which severely affect long bones length and is classified into type Ⅰ and Ⅱ[1,3]. Inheritance of type Ⅰ (Parenti – Franccaro) is autosomal recessive and characterized by poor ossification of the skull, pelvis

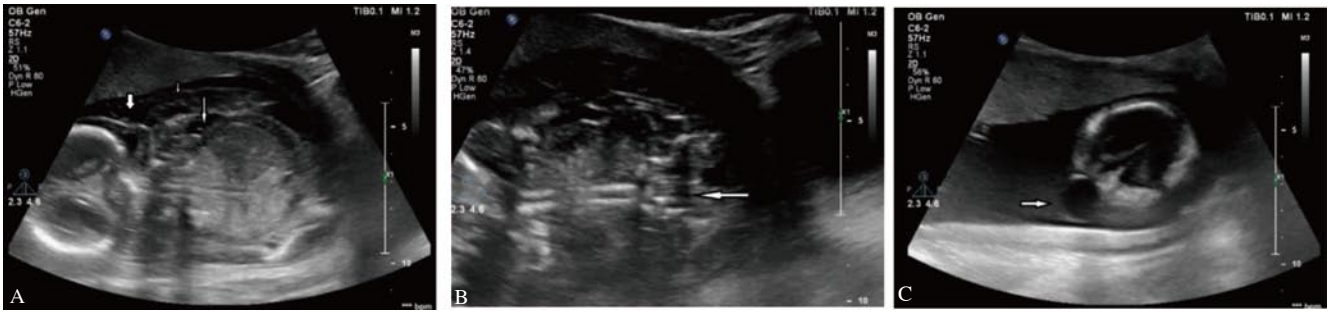

Figure 1. Ultrasonography at (17+4) wk.
(A) Extreme micromelia (short arrow), bell shaped thorax (long arrow), prominent abdomen and cystic hygroma (wide arrow); (B) Delayed ossification of sacral bones (arrow); (C) Good ossification off skull and occipital cephalocele (arrow).

and spine with rib fractures[1,3]. Type Ⅱ (Longer – Saldino) is usually caused by new autosomal dominant mutations in *COL 2A1* gene. Type Ⅱ is characterized by normal appearance of calvarium, absence of rib fracture and deficient ossification of spine (especially cervical and sacrum)[3-5]. Polyhydramnios and hydropic feature are also common as our case[1,3]. Pattern of deficient ossification and presence of rib fracture are good clues in differentiating between type Ⅰ and Ⅱ. Normal appearing calvarium and absence of rib fracture are in favor of type Ⅱ and vice versa[1]. Achondrogenesis can be differentiated from other lethal type skeletal dysplasia based on its features. In thanatophoric dysplasia, there is no deficient mineralization. The limbs are longer, and the thorax is narrow but is elongated in shape. Osteogenesis imperfecta is characterized by poor mineralization in multiple bones and multiple fractures are nearly hallmark of this dysplasia. Limbs are not as short as anchondrogenesis and head is not disproportionately large. Hypophosphatasia is characterized by diffuse hypo mineralization involving nearly all bones except clavicle. Micromelia is less severe comparing to achodrogenesis and there is no macrocrania[1,3].

Nearly all differentiating features and prognostic appearances can be evaluated by detailed anomaly scan by an expert, and radiography is nearly pathognomonic confirming option. In less severe and challenging cases, histopathology and gene study are helpful[1,4]. In our case, a high occiput cephalocele was present, and to the best of our knowledge, this is the second case with association of cephalocele and achondrogenesis type Ⅱ[6]. Lethality can be evaluated based on literature by thorax abdominal circumference ratio of <0.6 or FL to abdominal circumference <0.16 (which is severely affected in our case) in the presence of hydrops feralis[7]. Long bones were extremely short in our case, which were hardly visible (FL/foot: 0.02) and caused misdiagnosis of tetra phocomelia in other center.

In conclusion, we have described a case of achondrogenesis type Ⅱ with extreme micromelia, high occipital cephalocele, cystic hygroma and hydrops. Prenatal ultrasonography combined with post mortem radiography helped to reach the diagnosis.

Conflict of interest statement

The authors declare that they have no conflict of interest.

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