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◇ CASE REPORT ◇

Three-dimensional computed tomography complements ultrasonography in prenatal diagnosis of Pfeiffer type 2 syndrome: a case report

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Abstract

Pfeiffer syndrome—which is characterized by craniosynostosis, mid-face hypoplasia, and broad deviated thumbs—is classified into three clinical subtypes based on the severity of the phenotype. Pfeiffer syndrome type 2 is the most severe form, usually leading to death during early infancy. Therefore, an accurate diagnosis of Pfeiffer syndrome and appropriate counseling are critical. However, diagnosing Pfeiffer syndrome prenatally using ultrasonography alone is difficult because the differential diagnosis is complicated. Here, we report a case of Pfeiffer syndrome type 2 diagnosed in the second trimester using three-dimensional computed tomography complemented with ultrasonography. This report discusses the usefulness of computed tomography for the prenatal diagnosis of Pfeiffer syndrome.

Keywords

Pfeiffer syndrome, prenatal diagnosis, ultrasound, three-dimensional computed tomography, craniosynostosis

1. Introduction

Pfeiffer syndrome（PS）is an autosomal dominant disorder caused by mutations in the FGFR 1 or FGFR 2 gene and is characterized by craniosynostosis, mid-face hypoplasia, and broad and deviated thumbs[1]. This syndrome is very rare, affecting approximately 1 / 100, 000 live-born neonates, and is classified into three clinical subtypes based on the severity of the phenotype[1].

PS type 1 is characterized by mild manifestations, including brachycephaly, mid-face hypoplasia, normal neurological and intellectual development, and a good outcome[2]. However, PS type 2（PS 2）is the most severe form, presenting with a cloverleaf skull, severe ocular proptosis, elbow ankyloses, and large halluces and thumbs; affected individuals usually die in early infancy because of severe neurological compromise and respiratory problems[2]. Therefore, accurately diagnosing the type of Pfeiffer syndrome and appropriately counseling the family are critical.

The differential diagnosis is complicated because PS has wide clinical variability, and different causes of craniosynostosis have overlapping features[3]. Diagnosis is commonly made using ultrasonography（US）. However, the quality of the images depends on the sonographer's skills, fetal position, and size. Therefore, it could be difficult to diagnose in detail prenatally using US alone. The usefulness and safety of three-dimensional computed tomography（3D-CT）have recently been reported for the prenatal diagnosis of fetal skeletal anomalies, especially fetal skeletal dysplasia[4]. Here, we report a case of PS 2 diagnosed in the second trimester using 3D-CT complemented with US.

2. Case presentation

A healthy 41-year-old nulliparous Japanese woman underwent a fetal US scan at 20 weeks of gestation, which showed scalloping of the frontal bones（**Fig. 1**）.

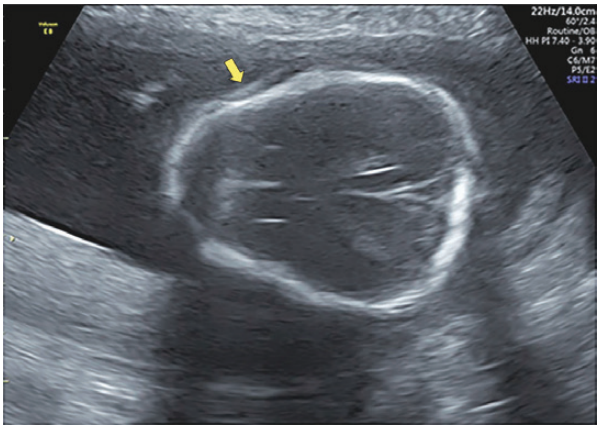

Fig. 1 An ultrasonographic scan at 20 weeks of gestation showed scalloping of the frontal bones（arrow）, which resembled the lemon sign.

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