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产前超声诊断胎儿全前脑及其面部畸形

Prenatal ultrasonic diagnosis of fetal holoprosencephaly and facial anomalies

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中文摘要:

目的 观察胎儿全前脑畸形(HPE)及其面部畸形的声像图特点,探讨产前超声诊断HPE的价值。方法 回顾性分析我院产前超声诊断或拟诊的15胎HPE的超声表现,与引产后尸体解剖结果进行对照分析。结果 15胎HPE中,无叶型10胎,半叶型5胎,均伴不同程度、不同部位面部畸形,其中独眼畸形7胎、正中唇腭裂4胎、猴头畸形2胎、头发育不全畸胎1胎、轻度眼距过近1胎。27处面部畸形中,以眼、鼻、唇、腭等中轴结构多处受累、多种畸形组合为主,超声诊断符合率为70.37%(19/27)。10胎合并面部以外其他结构异常(共30处),7胎为多发畸形,超声诊断符合率76.67%(23/30)。结论 超声可准确显示HPE的典型颅内改变和特殊的面部畸形,有助于诊断及鉴别诊断。

英文摘要:

Objective To investigate ultrasonic features of fetal holoprosencephaly (HPE) and facial anomalies, and to explore the value of prenatal ultrasonography in diagnosis of HPE. Methods The ultrasonic manifestations of 15 fetuses with HPE diagnosed or suspected with prenatal ultrasound were retrospectively analyzed and compared to the autopsy results. Results There were 10 fetuses of alobar and 5 of semilobar. Facial defects with different degrees and different positions were found in all 15 fetuses, including 7 fetuses with cyclopia, 4 with middle cleft of lip and palate, 2 with cebocephaly, 1 with ethmocephaly and 1 with milder hypotelorism. There were totally 27 facial anomalies mainly involving midline structures such as eyes, nose, lip and palate, characterized by multiple combinations of multi-deformations. The diagnostic accordance rate of prenatal ultrasound for 27 facial anomalies in HPE fetuses was 70.37% (19/27). Extrafacial malformations were found in 10 fetuses (30 lesions), and in 7 fetuses multiple malformations were noticed, and the diagnostic accordance rate of ultrasound for extra-facial malformations of HPE was 76.67% (23/30). Conclusion Classic intracranial abnormalities and characteristic facial features of HPE could be demonstrated accurately with prenatal ultrasound, which is helpful to the diagnosis and differential diagnosis of HPE.

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