대한산부인과초음파학회추계학술대회초록작성지

구연( ), 포스터(∨)

삼중검사 이상과 초음파상 태아의 구조적 이상이 동반되어 임신 중반기에 진단된 complete trisomy 9

정유아, 조금준, 김정숙†, 박선화†, 오민정
고려대학교 의과대학 산부인과학교실, †유전병연구소

ABSTRACT

Second-trimester diagnosis of complete trisomy 9 associated with abnormal maternal serum screen results and sonographically detected structural abnormalities

Yu Ah Jeong, M.D., Geum Joon Cho, M.D., Jung Sook Kim†,
Sun Hwa Park, M.D.†, Min Jeong Oh, M.D.
Department of Obstetrics and Gynecology, †Institute of Human Genetics,
Korea University School of Medicine

Trisomy 9, mosaic or nonmosaic, is a relatively rare chromosomal abnormality, comprising only 2.7% of all trisomic cases. Since the vast majority of affected fetuses are miscarried spontaneously in the first trimester, trisomy 9 is rarely seen thereafter, there is a paucity of reports regarding the prenatal sonographic findings of trisomy 9. Clinical and sonographic findings that have been described include intrauterine growth restriction, central nervous system abnormalities, cranial and facial anomalies, skeletal defects, congenital heart defects, and urogenital abnormalities.

We report a fetus with sonographically detected structural abnormalities and abnormal maternal serum screen results showing an elevated maternal serum alpha-fetoprotein (MSAFP) level and low maternal serum free β-human chorionic gonadotrophin (MSfreeβ-hCG) level which was subsequently found to have a complete trisomy.