عنوان مقاله:

Prenatal Diagnosis for Abnormal Sonography Suggestive of Chromosome Abnormality

محل انتشار:

سومین کنگره بین المللی و پانزدهمین کنگره ملی ژنتیک ایران (سال: 1397)

تعداد صفحات اصل مقاله: 1

نویسندگان:

f mousavi - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

I Bagherizadeh - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

E SAEDI - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

N Mohammadkhani - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

N Ghadami - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

A Abdi - *Department of Medical Genetics and Sarem Cell Research Center (SCRC), Sarem Women s' Hospital, Tehran, Iran*

خلاصه مقاله:

Background: The use of ultrasound in prenatal diagnosis of fetal genetic disorders is rapidly evolving. Use of three-dimensional and four-dimensional ultrasound as an adjunct to two-dimensional ultrasound in the diagnosis of fetal structural and chromosomal abnormalities is a developing modality. Most fetuses with cytogenetic abnormalities have either external or internal defects that can be recognized on detailed ultrasonography examination. Many of these defects are easily detected at routine examination, but others need to be looked for specifically. The commonest detectable chromosomal abnormalities are trisomy 18, followed by trisomy 21, triploidy, Turner syndrome, unbalanced chromosomal rearrangement, and trisomy 13. The ultrasonographically detectable phenotypic expression of the different types of chromosomal abnormalities could be useful in genotypic and phenotypic correlations. Methods and materials: In this study we report four patients with chromosome abnormalities following high risk abnormal sonography in the pregnant women referred for amniocentesis to Sarem Women's hospital in Tehran. They underwent amniocentesis for karyotyping using standard high resolution GTG banding technique. 15-50 metaphase spreads were studied using light microscope. Results: Karyotype of four fetuses with high risk abnormal sonography are as below: 47,XX,+mrdn; 47,XY,del(7)(p13p15)dn; 46,XX,t(3;17)(p21;q11.2)dn, and 45,X. Conclusion: These results demonstrate that presence of any kind of ultrasonography abnormality can significantly increase the risk of fetal chromosome abnormalities. It is therefore of paramount importance to carry out prenatal diagnosis using karyotyping in such cases.

کلمات کلیدی:
ultrasonography, prenatal diagnosis, amniocentesis