Abstract

Foetal anomalies found on ultrasound increase the probability of occurrence of chromosomal abnormalities. They cause about one quarter of all abortions and stillbirths and many of inborn defects in newborns.

Karyotype analysis is number one method in prenatal diagnosis whereas array CGH is often used as a verification and supplemental method. The aim of this work was to prove that array CGH gives additional chromosomal findings to karyotypes and could substitute conventional karyotyping as a primary examination method in foetuses with ultrasound findings.

We examined 45 prenatal samples using both methods. These samples were referred for invasive examination because of abnormal ultrasound findings. Karyotype analyses found two abnormalities in two (4,4 %) patients and array CGH identified aberrations in five (11,1 %) foetuses whereas both anomalies detected by karyotypes were discovered by array CGH too. This means that array CGH identified about 6,7 % more aberrations than karyotype.

Our results correspond with scientific articles which refer that array CGH should replace karyotype not only in postnatal examinations but even in prenatal diagnosis.

Keywords: chromosomal aberrations, array CGH, karyotype, prenatal diagnosis, ultrasound