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# Systematic review of accuracy of prenatal diagnosis for abnormal chromosome diseases by microarray technology

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**ABSTRACT.** The accuracy of prenatal diagnosis for abnormal chromosome diseases by chromosome microarray technology and karyotyping were compared. A literature search was carried out in the MEDLINE database with the keywords “chromosome” and “karyotype” and “genetic testing” and “prenatal diagnosis” and “oligonucleotide array sequence”. The studies obtained were filtered by using the QUADAS tool, and studies conforming to the quality standard were fully analyzed. There was one paper conforming to the QUADAS standards including 4406 gravidas with adaptability syndromes of prenatal diagnosis including elderly parturient women, abnormal structure by type-B ultrasound, and other abnormalities. Microarray technology yielded successful diagnoses in 4340 cases (98.8%), and there was no need for tissue culture in 87.9% of the samples. All aneuploids and non-parallel translocations in 4282 cases of non-chimera identified by karyotyping could be detected using microarray analysis technology, whereas parallel translocations and fetal triploids could not be detected by microarray analysis technology. In the samples with normal karyotyping results, type-B ultrasound showed that

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6% of chromosomal deficiencies or chromosome duplications could be detected by microarray technology, and the same abnormal chromosomes were detected in 1.7% of elderly parturient women and samples with positive serology screening results. In the prenatal diagnosis test, compared with karyotyping, microarray technology could identify the extra cell genetic information with clinical significance, aneuploids, and non-parallel translocations; however, its disadvantage is that it could not identify parallel translocations and triploids.

**Key words:** Prenatal diagnosis; Abnormal chromosome diseases; Microarray

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