MGZ

Medical Genetics Center

Clinician Information

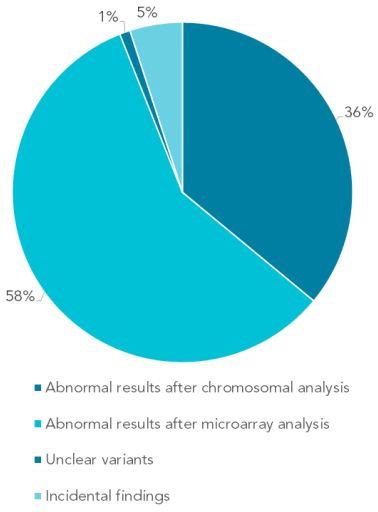
# PRENATAL CHROMOSOMAL MICROARRAY ANALYSIS

## PRENATAL CHROMOSOMAL MICROARRAY ANALYSIS (CMA)

**Prenatal CMA (molecular karyotyping)** uses modern molecular-biological techniques that enable chromosomal analysis of the unborn child's DNA with a very high resolution.

Chromosomal microarray analysis (CMA) is chip-based karyotyping that is conducted from genomic DNA and does not require metaphase chromosomes. It has a resolution that is approximately one thousand times higher than that of conventional chromosome analysis, thus making it possible to detect unbalanced chromosomal aberrations which are only a few kilobases in size. MGZ uses a single nucleotide polymorphism (SNP)-based microarray chip (Infinium CytoSNP 850K, Illumina).

- A recent study confirmed the diagnostic superiority of prenatal CMA compared to conventional chromosomal analysis and NIPT (Vogel et al., Ultrasound Obstet Gynecol, 2018). In particular, NIPT and chromosomal analysis are often unable to detect the vast majority of chromosomal abnormalities in patients with a moderately increased risk (1:50 to 1:300) after first trimester testing.
- Similar results were reported by Vogel et al. following a study on patients with conspicuous ultrasound findings. Thirty-six percent of the fetuses showed numerical chromosomal abnormalities, which were detected through conventional analysis. Fifty-eight percent showed chromosomal abnormalities, which were only detected through microarray analysis. In only one percent of cases, the results could not be clearly classified. In five percent, incidental findings were observed that were not associated with the conspicuous ultrasound findings (Figure 1).



## Advantages of Prenatal CMA

- Objective, high-resolution depiction of the fetal karyotype
- Resolution up to 1000 times higher than light microscopic analysis
- Detection of maternal contamination
- Simultaneous testing for all microdeletion / microduplication syndromes
- Significant diagnostic gain compared to light microscopic analysis
- Detection of genomic dosage changes at exon level

## Indications for Prenatal CMA

- Chromosomal aberration detected through light microscopy, which cannot be characterized clearly: In such cases, additional microarray analysis should be recommended for differentiated examination of structural chromosomal aberrations
- It is not unusual to have fetal abnormalities that cannot be attributed to any numerical chromosomal defects or that may be associated with small structural chromosomal aberrations, which are not detected through conventional cytogenetic analysis.
- **Data from international studies show that invasive diagnostics and prenatal microarray analysis are the best diagnostic options, particularly for women who are categorized in the medium risk range based on the results of non-invasive diagnostic testing (increased nuchal translucency, irregular first trimester screening).**

## Limitations of CMA

Despite the ability to deliver much higher informative value, prenatal CMA cannot detect all causes of genetic diseases.

For instance, no conclusion can be made in the case of monogenic diseases that are caused by a pathological change in the sequence of individual genes. In cases of suspicion from the examination results or from the family history, a targeted molecular-genetic analysis is necessary.

## Price Inquiries

A cost statement can be requested by sending an email to inquiry@mgz-muenchen.com or by using our convenient contact form found under Cost & Billing Information at www.mgz-muenchen.com.

On our website, you will find information on other clinical topics as well as information about our company and services.

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