Prenatal Microarray
also known as: Prenatal Array, CGH

TEST DESCRIPTION:
As opposed to screening tests, Prenatal Microarray can provide diagnostic insight for current pregnancies. This test detects clinically significant large (whole chromosome) and submicroscopic (microdeletion/microduplication) copy number changes throughout the genome in fetal cells.

TEST DETAILS and COMPLEMENTARY TESTING:
- This test can be completed concurrently with Prenatal Chromosome Analysis or as a reflex test if Chromosome Analysis does not identify an abnormality.
- In conjunction with Chromosome Analysis, FISH, and/or Microarray, Amniotic Fluid AFP (AF-AFP) can be ordered on amniotic fluid samples with a gestational age of 22 weeks 6 days or less. (AChE should be ordered, instead of AF-AFP, if gestational age is 23 weeks or greater). In most situations if AF-AFP is positive, AChE will be completed. These non-genetic send-out tests can be ordered on our laboratory’s test request form, but are performed, reported, and billed by UNMC Regional Pathology Services and questions for these tests should be directed to their laboratory by calling 402-559-6420.
- Pregnancy Loss Microarray should be ordered instead of this test to analyze cells in a tissue sample after a pregnancy loss.
- Postnatal testing on maternal, paternal, or newborn blood after delivery, including Postnatal Chromosome Analysis, Postnatal FISH, or Postnatal Microarray, must be ordered separately.

ADVANTAGES:
- Evaluates hundreds of genetic conditions across the genome with one test
- Detects aneuploidy (including trisomy and sex chromosome abnormalities) and triploidy
- Identifies sub-microscopic deletions and duplications in regions known to be associated with well-characterized microdeletion and microduplication syndromes
- Provides enriched coverage of subtelomeric regions, often undetectable by traditional chromosome analysis
- More comprehensive than individual FISH tests
- Circumvents cell culture in most cases
- Often allows for results on suboptimal specimens for which chromosome analysis is not feasible

LIMITATIONS:
- Cannot identify all genetic conditions or the cause of all fetal anomalies in a pregnancy
- Does not detect changes in the DNA sequence of genes
- Cannot detect balanced chromosome rearrangements (a complementary test such as Prenatal Chromosome Analysis should be performed to detect balanced chromosome rearrangements)

INDICATIONS FOR TESTING: Microarray has been recommended by the American College of Obstetrics and Gynecology (ACOG) when fetal ultrasound anomalies are detected or when a patient is undergoing invasive prenatal diagnostic testing. This test may be particularly beneficial to patients with the following clinical indications:
- Abnormal ultrasound findings
- Abnormal screening tests
- Family history of a genetic or chromosomal abnormality that is detectable by microarray technology
- History of pregnancy loss

Access our Prenatal Microarray Disorders List on our website.
SPECIMEN COLLECTION & TRANSPORT:
Complimentary test kits are available upon request, but are not required.

SAMPLE TYPE and REQUIREMENTS:
- **amniotic fluid:** 15 – 20 ml in sterile tube(s)
  - Additional specimen is required to perform Cytogenetic Analysis and/or FISH. Refer to our Prenatal and Pregnancy Loss specimen requirements for more details.
  - When Prenatal Microarray is ordered with maternal cell contamination analysis, 2 – 5 ml blood EDTA is also required.
- **chorionic villi sampling (CVS):** 10-20 mg in tissue culture media
- **fetal fluid / urine:** 15 – 20 ml in sterile tube(s)

SHIPPING:
- Maintain and ship samples at room temperature.
- Coordinate transport for sample to be received in our laboratory within 24-48 hours of collection.
  - **LOCAL:** Call 402-559-5070 (option 1)
  - **OUT OF AREA:** Prior to shipment, please fax the completed test request form to 402-559-7248, including the FedEx® airbill tracking number.
    - Saturday delivery MUST be checked when sending FedEx® on Friday.
    - Please include Internal Billing Reference # 3155070600 on the FedEx® airbill.
    - **Ship To:** Human Genetics Laboratory – Zip 5440
      UNMC Shipping & Receiving Dock
      601 S. Saddle Creek Road
      Omaha, NE 68106

FORMS FOR TESTING: The following forms can be downloaded via our website.
- **Required:** Prenatal and Pregnancy Loss Test Request Form
- **Optional:** Informed Consent for Genetic Testing

POTENTIAL TEST RESULTS:
NORMAL RESULTS:
- A normal result indicates no clinically-significant chromosome abnormalities were identified.
  - arr[hg19](1-22,X)x2 (female)
  - arr[hg19](1-22)x2,(XY)x1 (male)

ABNORMAL RESULTS:
- Results are reported by location in the genome, including chromosome and size.
- **Deletion:** Part of a chromosome (genetic material) is missing. Some may be very small and only include one gene and others are bigger and may involve numerous genes.
- **Duplication:** Extra chromosome material is present in the patient’s DNA.
- An abnormal result indicates that a clinically-significant chromosome abnormality was identified in the fetus that likely provides an explanation for the indications.
  - **Pathogenic** – Reported when a copy number variant (CNV) is associated with a known syndrome (see Prenatal Disorders List) that is covered by this array.
  - **Uncertain Clinical Significance (UCS)** – Our Prenatal Microarray is designed to identify clinically-relevant anomalies. However, in rare cases, a reportable anomaly may be identified for which a definitive classification (i.e., normal or abnormal) is not possible. In those cases, these anomalies will be reported as variants of uncertain clinical significance. Uncertain variants may also be classified as “Likely Pathogenic” or “Likely Benign” based on the ACMG recommendations for variant classification.2
- Parental testing (Postnatal Chromosome Analysis on maternal and/or paternal blood) may be recommended in order to clarify whether the result is de novo or familial for the purpose of recurrence risk calculation.

TURNAROUND TIME: Results are typically available in 1 - 2 weeks on all specimen types.
BILLING: Our laboratory offers patient/self-pay, insurance (including Medicare/Medicaid), and client/institution billing options. Verifying coverage requirements or obtaining preauthorization PRIOR TO OR AT THE TIME OF SPECIMEN COLLECTION is often necessary. We provide preauthorization services upon request by calling 402-559-5070 (option 3); the following form is helpful for obtaining the information required by insurance providers and can be downloaded via our website.

- Insurance Preauthorization Request

In some circumstances, a test may be warranted even though insurance coverage is denied or not guaranteed. For these situations, we request the following form be signed by the patient and submitted with the sample. This helps inform patients of their potential financial responsibility, should the costs of genetic testing not be paid by their insurance provider.

- Advanced Beneficiary Notice of Noncoverage (ABN) – required when billing Medicare

CPT CODES:

- amniotic fluid, CVS, fetal fluid/urine: 81229, 88235

PRICING: For current costs contact the laboratory billing staff at 402-559-5070 (option 3).

RESOURCES: The following resources can be downloaded via our website, or you may request brochures for your clinic by contacting our marketing specialist at 402-559-6935 | humangenetics@unmc.edu.

- PATIENT BROCHURE: Amniocentesis (English)
- PATIENT BROCHURE: Amniocentesis (Spanish)
- PATIENT BROCHURE: CVS (English)
- PATIENT BROCHURE: CVS (Spanish)

REFERENCES:


- Prenatal Disorder List

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