Chromosomal Analysis in Prenatal Patients

Description

Chromosome analysis is a study of the number and general structure of all 46 chromosomes; it is also known as a karyotype. In a standard karyotype, chromosomes from cultured cells in the body (usually white blood cells) are counted to ensure that the cells evaluated have the correct number of chromosomes, and their structure is evaluated to ensure that there are no large pieces of material that are missing (deleted), extra (duplicated), or rearranged in any way.[1]

Chromosomal microarray (CMA) uses molecular techniques to examine chromosomes for copy number changes (deletions or duplications) that are 5-10 times smaller than can be detected by a standard karyotype. In contrast to a karyotype, CMA does not require cells to be actively dividing, which can be useful for some prenatal indications.

Recommendations[2]

1. It is **not appropriate** to utilize Karyotype or CMA as an initial screening test for a routine pregnancy or for the evaluation of the first or second occurrence of a first trimester miscarriage or first unexplained second trimester pregnancy loss.
2. It may be appropriate to order chromosomal analysis when one of the following indications is present:
    a. A history of two or more previous first trimester or one previous second trimester unexplained pregnancy loss defined as a patient who has undergone a full evaluation for pregnancy loss and for which no cause has been found.
    b. Documentation of an abnormal fetus in the current pregnancy (maternal serum screen, abnormal sonogram, lesions identified at autopsy, etc.)
3. At present, a Karyotype should be the first test performed, and CMA reserved for the following circumstances:
    a. Karyotype returns a normal result
    b. The tissue submitted for Karyotype fails to grow (30% of cases)
    c. The tissue available for Karyotype is highly unlikely to grow (e.g. macerated specimen).

Resources:

1. Standard Chromosome Analysis
   http://genetics.emory.edu/pdf/Emory_Human_Genetics_Chromosome_Analysis.PDF
2. Consensus recommendations from Intermountain Maternal-Fetal Medicine and the Genetic Testing Practice Council.