Chromosome Testing During Pregnancy

KARYOTYPE or CHROMOSOMAL MICROARRAY

Prenatal testing can detect chromosome conditions in a baby before birth. You can decide which chromosome test is right for you.

What is a standard chromosome study?

A standard chromosome study is called a karyotype. A karyotype counts the number of chromosomes in each cell and examines the way each chromosome looks under a microscope. This test accurately detects common chromosome conditions, like Down syndrome. These conditions are more likely to happen as a woman gets older. Results from a karyotype take about two weeks. Some small changes in the chromosomes cannot be found by a karyotype.

Did you know?

- Small changes in the chromosomes can happen in any pregnancy
- Some chromosome conditions are not related a woman's age
- Not all changes in the chromosomes will cause health problems in a baby
- These tests do not detect every genetic disease or chromosome condition.

What is a chromosomal microarray?

Chromosomal microarray (or "microarray") detects missing or extra pieces of chromosomes, including very small changes called micro-deletions and micro-duplications. These type of small chromosome changes happen in less than 1 in 100 (1%) pregnancies and are not related to the age of the mother. A baby is more likely to have a micro-deletion or micro-duplication when there are ultrasound findings. Microarray testing will find common chromosome conditions, like Down syndrome, and can also find chromosome conditions that would not be seen with a karyotype. Microarray results are usually available within 2 weeks. A small number of samples take longer (up to 3 weeks).

Why would I choose microarray testing?

- Microarray can find more chromosome conditions during pregnancy than a karyotype.

Why would I not choose microarray testing?

- Microarray is more likely to find a ***variant of unknown significance (VUS)***.

  About 1 in 20 (5%) microarray results find a VUS. This is when a chromosome change is found, but there is not enough information to know if it will cause any problems. This type of result may cause anxiety for expectant parents and can make it hard to decide the best way to manage your pregnancy.

- Some ultrasound findings may suggest a rare condition that is not detected by microarray.

  Ultrasound findings can let your provider know if a different test would be recommended. These conditions are not related to the mother's age.

Which chromosome testing will be done for my pregnancy?

The decision about which chromosome testing to have is up to you. Either test will accurately diagnose chromosome conditions related to age, like Down syndrome.

Microarray can give you extra information about uncommon chromosome conditions, but there is a higher chance of getting a variant of unknown significance.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional.

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