UW Medicine
UNIVERSITY OF WASHINGTON MEDICAL CENTER

Preimplantation Genetic Testing

Testing embryos before they are placed in the uterus

This handout explains preimplantation genetic testing, how the tests are done, and what to expect.

What is preimplantation genetic testing?

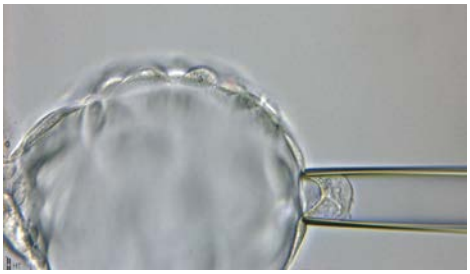
Embryos that are formed as part of an *in vitro fertilization* (IVF) process can be tested for abnormal chromosomes. We do this testing before the embryos are placed in your uterus.

This testing can be helpful because many embryos with abnormal chromosomes may not bring about pregnancy. If they do result in pregnancy, there is a higher risk of miscarriage, birth defects, or other health problems.

With *preimplantation genetic testing*, you can choose to have only embryos with normal chromosomes placed in your uterus.

What types of tests are done?

There are 2 types of tests we can do on embryos before they are placed in your uterus: *preimplantation genetic testing for aneuploidy* (PGT-A) and *preimplantation genetic testing for monogenic (single gene) disorders* (PGT-M).



At the start of the test, a few embryo cells are gently pulled into a collection tube.

The cells that are removed from the embryo are then sent to the lab for genetic testing.

Preimplantation Genetic Testing for Aneuploidy

PGT-A tests an embryo for these abnormalities:

- **Too few or too many chromosomes.** There are 46 chromosomes in a normal embryo. If an embryo has more or less than this number, the baby may have Down syndrome (Trisomy 21), Trisomy 13, or other chromosomal abnormalities.

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- **Chromosomes in the wrong order**. If the normal 46 chromosomes are not in the correct order, it is called a *translocation*. Translocations can cause problems with growth or health of a child.

Anyone having IVF can choose to do PGT-A. But we often strongly advise the test if you:

- Have had more than 1 miscarriage;
- Are at least 37 years old;
- Or have a smaller supply of eggs (smaller *ovarian reserve*)

Preimplantation Genetic Testing for Monogenic Disorders

PGT-M tests an embryo for a specific genetic disorder. PGT-M is done for prospective parents who have a known genetic disorder or are carriers of a disorder. PGT-M helps identify embryos that do **not** have that specific genetic disorder.

How is this testing done?

- Cells from each embryo are removed and sent for genetic testing. This process is called a *biopsy*.
- Different methods are used for this genetic testing, depending on the embryo's stage of growth.

What can I expect if I decide to have these tests?

- Embryos that undergo these tests must be frozen while the testing takes place.
- You will later undergo a *frozen embryo transfer* (FET) cycle to prepare your uterus. Then, one or more embryos will be placed in your uterus to bring about possible pregnancy. To learn more about FET, ask your provider at University Reproductive Care (URC) for the handout "Frozen Embryo Transfer Cycle."

What risks are involved?

- Rarely, the biopsy can damage the embryo.
- It is possible to get no result for a tested embryo.
- The test results are not 100% accurate. Please talk with your URC provider about all your prenatal genetic testing options during pregnancy.

Questions?

Your questions are important. Call your doctor or healthcare provider if you have questions or concerns.

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