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 screening (PGS) and preimplantation genetic diagnosis (PGD).

Preimplantation Genetic Screening

PGS 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.

- **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.

Talk with your provider if you have any questions about preimplantation genetic testing.
Anyone undergoing IVF can choose to do PGS. But the test is often strongly advised for a woman who has had more than one miscarriage, is older, or has a smaller supply of eggs (ovarian reserve).

**Preimplantation Genetic Diagnosis**

PGD tests an embryo for a specific genetic disorder. PGD is done for prospective parents who have a known genetic disorder or are carriers of a disorder. PGD 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.

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**Questions?**

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

University Reproductive Care: 206.598.4225

[www.uwmedicine.org/uwfertility](http://www.uwmedicine.org/uwfertility)