**GENETIC TESTING**

As the cost of genetic analysis decreases and as research advances, it is becoming increasingly possible to include a person’s genetic makeup in the repertoire of tools that inform their health and well-being. Similarly, genetic analysis can be used in reproductive healthcare to learn about the DNA of an embryo in a Petri dish or a fetus in the womb, often as a way to gain medical insights at early stages of development. Two such genetic technologies are: **preimplantation genetic diagnosis (PGD)** and **non-invasive prenatal testing (NIPT)**, which allow people to screen embryos and fetuses, respectively, for a variety of characteristics.

**Prenatal testing**

Technology is transforming the ways that a person could learn about the genetic makeup of the fetus they are carrying. Previously, the only way doctors could analyze the DNA of a fetus was through an invasive procedure, either amniocentesis (typically at 15-20 weeks of pregnancy) or chorionic villus sampling (CVS; typically at 11-13 weeks of pregnancy). Since these invasive procedures involve collecting tissue or fluid from inside the womb, they both carry a small risk (how small remains a matter of debate, but could be anywhere from a .005% to 1% chance) of miscarriage. In 2011, a new generation of non-invasive prenatal tests (NIPT) became available for analyzing fetal DNA through a blood sample taken from a pregnant person’s arm. NIPT can be performed as early as 9 weeks of pregnancy and, as with any blood test performed during pregnancy, NIPT does not increase the risk for miscarriage.

NIPT is most commonly used to screen for extra or missing copies of certain chromosomes, that can result in conditions such as Down Syndrome. NIPT can also assess the chromosomal sex of the fetus. This test is more accurate than previous generations of prenatal blood tests, which did not look directly at fetal DNA. Importantly, NIPT is not diagnostic. Because NIPT can be performed at an early stage of pregnancy and only requires a blood sample, it has been rapidly adopted by medical professionals and raises challenges around informed consent. As a result, people are grappling with how to handle genetic information about the developing fetus that is increasingly available to prospective parents. Ethical and practical questions abound about how this information might be used. These questions include (i) whether information learned via NIPT could improve medical care; (ii) how NIPT could impact pregnancy termination rates; and (iii) whether the availability of NIPT might add to the stigmatization of people with perceived disabilities.

**Embryo testing**

Preimplantation genetic diagnosis (PGD) allows for the genetic diagnosis of embryos created by in vitro fertilization (IVF) - the fusion of egg and sperm in a lab. Based on the results of this analysis, one or more embryos can be selected for transfer into the womb. PGD can be used to assess whether an embryo has genetic variants that are associated with fatal diseases, such as Tay-Sachs disease and Huntington’s Disease, with the goal of avoiding them. Thousands of healthy children have been born as a result of this technology, free of the genetic diseases that have, in many cases, devastated the older generations of their families. At the same time, PGD has raised ethical issues, as it gives individuals the capacity to select one embryo over another, and therefore brings to the forefront issues about autonomy, medical interventions, and disability.

**GENE THERAPY**

Gene therapy is an experimental technique that uses genes to treat or prevent disease. In the future, this technique may allow doctors to treat a disorder by inserting a gene into a patient’s cells instead of using drugs or surgery. Researchers are testing several approaches to gene therapy, including:

* Replacing a mutated gene that causes disease with a healthy copy of the gene.
* Inactivating, or “knocking out,” a mutated gene that is functioning improperly.
* Introducing a new gene into the body to help fight a disease.

Although gene therapy is a promising treatment option for a number of diseases (including inherited disorders, some types of cancer, and certain viral infections), the technique remains risky and is still under study to make sure that it will be safe and effective. Gene therapy is currently being tested only for diseases that have no other cures.