



PGD: Fact vs. Fiction. Genesis Genetics Institute sets the record straight about PGD and its uses in HLA matching

Preimplantation Genetic Diagnosis (PGD) is in the news this week more than ever due to the release of the cinema blockbuster, *My Sister's Keeper*. The scientists at Genesis Genetics Institute were the first to pioneer and successfully use this testing method for identifying embryos affected by a specific genetic disease and recognize the importance of clearly explaining the capabilities and limitations of PGD in terms of the movie's storyline.

PGD had its beginnings twenty years ago, according to Dr. Mark Hughes, President of Genesis Genetics Institute, "Couples would come to see me after losing a baby to a terrible inherited disease, and they would ask, 'What will happen if we try to get pregnant again?' I would look them in the eye and tell them that they were rolling loaded genetic dice, and that they had a 25% chance (or more) of having yet another baby with the same disease. Their options were: 1) not have any more children since the risk is so high; 2) adopt; or 3) get pregnant again and hope for the best. It seemed like there must be a better alternative to help these desperate couples build their families. We realized that *in vitro* fertilization (IVF) was being routinely used to for infertile couples, and that it should be possible to test an IVF-produced egg/embryo before it was ever placed into the womb. The idea was to make the genetic diagnosis before a pregnancy ever began. Surely, this was a better alternative to conventional prenatal testing with an amniocentesis. The couple could begin their pregnancy, on day one, knowing that the genetic disease in their family was avoided. Effectively this would eliminate the disease from their family tree forever."

This technology is performed many times every day, in conjunction with hundreds of IVF clinics around the world. Today, the Genesis Genetics Institute tests for over than 300 inherited diseases. The couple visits their local doctor and undergoes standard IVF as though they were infertile – although usually are not. A test is performed on the fertilized eggs that they produce, and healthy ones are transferred to the uterus.

Over ten years ago, Dr. Hughes and his team were approached by a couple with a child afflicted with a life threatening, incurable disease that could be cured only with an "identical" sibling cord blood transplant. They wanted additional children as well and asked if it might be possible to test their fertilized eggs to avoid having yet another child with this disease (one-in-four chance). They also wondered if we could test, at the same time, for those IVF-embryos that were also a perfect sibling "HLA" transplant match to their sick older daughter. Indeed, the couple went on to have a healthy son. At birth, his umbilical cord blood, loaded with stem cells, was saved and later given to his sister. "In effect," Dr. Hughes said, "we were able to hand this couple a healthy baby and save the life of the sibling". The British call this technology "Savior Sibling", and it has now been performed many times for a wide range of diseases. It can also be used for some cancers such as leukemia. And, this raises ethical and socially provocative issues.

While this is the premise behind the film "*My Sister's Keeper*" it is important to note the difference between real science and a fictional story. In the film, the second child was put through years of medical tests and procedures. Her blood, marrow and other cells were used year after year to keep her older sister alive. Later, there was discussion about taking a kidney from the younger sibling.



None of this is remotely close to reality. Hughes explains, "If PGD for HLA matching is used to select an embryo, then the cord blood or bone marrow collected at birth is used just once to CURE the sibling's disease. This has been used successfully (many times) for sickle cell anemia, Beta-Thalassemia, Diamond-Blackfan and Fanconi anemias and numerous other conditions. It has also been used in the setting of leukemia. But, the literary license used in the film has never occurred in over 15 years of performing this technology. When the new baby is born, the umbilical cord blood is collected as a one-time event, with the end result being two healthy children. In no way is PGD used to produce a sibling who could be depended upon for years to come to keep an older sibling alive. And, this technology has never been used (nor asked by anyone to be used) for solid organ transplantation, which would be irresponsible and unethical". \

We welcome your questions about PGD and PGD to find an HLA match. Feel free to email us at counselor@genesisgenetics.org.