Health Information Technology/Trend

Lynette Barnhart, Shayna Bauman, Rosa Carranza, Marissa Hampton, Magidah Kobty

University of Texas Medical Branch at Galveston School of Nursing

Informatics and Quality Improvement

GNRS 5349

Dr. Karen O’Brien, PhD. ACNP-BC

June 10, 2013
Introduction

The study of the Human Genome Project, has granted scientists and researchers unprecedented access to the genetic blueprint of human beings. As a result of human genome discoveries, great advances have been made in the fields of genetics and genetic technology (United States Department of Energy Genome Programs, 2011). A growing trend is emerging among parents who wish to use genetic screening technology for non-medical selection of genetic traits, such as gender. This phenomenon has given rise to the near future prospect of producing “designer babies” whose genetic makeup has been modified to produce pre-selected, desired characteristics (Steinbock, 2008). The increasing use of technologically assisted reproduction, genetic screening, and diagnosis has created new ethical debates, implications for clinical nursing practice and education, as well as patient education needs. Additionally, healthcare system analysis will need to examine the availability, cost, and interoperability of these procedures. Nurses, Nurse Practitioners (NPs), and other healthcare providers working in the areas of fertility, genetics, women’s health, and neonatology must possess a thorough understanding of these growing trends, procedures involved with the use of this medical technology, and the educational and emotional needs of recipients in order to support and provide the best care for these couples and their offspring (Hershberger, Shoenfeld & Tur-Kaspa, 2011).

Descriptions of Technology/Trend

Pre-Implantation Genetic Diagnosis (PGD) was first introduced in the 1990s in order to aid couples who were at high risk for x linked genetic diseases. As a result only female embryos were transferred via invitro fertilization (IVF), eliminating the possibility of inherited x linked
diseases (Sermon, K., Van Steirteghem A., & Liebaers, I., 2004). Furthermore, Pre-Implantation Genetic Screening (PGS) methods consist of examining embryos, from parents whom are presumed to be chromosomally normal, for aneuploidy (Dayal, 2011).

Companies like The Fertility Institutes offer both PGD and PGS services to their customers (“Sex selecting, 2001-2013”). The company screens and eliminates embryos with genetic abnormalities and/or diseases prior to IVF. This type of preconception testing replaces post conception procedures such as amniocentesis and chorionic villus sampling, where genetic abnormalities are recognized and then the decision to terminate the pregnancy is presented (Dayal, 2011). According to Dayal (2011), there is no increase in fetal malformation rates or other problems in relation to PGD.

Currently through PGD and PGS companies have taken screening to the next level. In addition to aneuploidy screening, gender selection services are an option available to customers. The Fertility Institute predicts that couples in the future will have the ability to screen their embryos for traits such as hair color, eye color, and complexion (Hub, 2009). This technology is limited by the DNA of the donating couple; traits are only possible if the parents have that particular gene (Hub, 2009).

Analysis

As with any new technology or trend there is an impact on nursing knowledge, nursing practice, access to care, patient centered care, patient safety, quality of care, cost, ease of use, interoperability, availability, education/training needs of staff or patients, potential ethical issues, privacy issues, and regulatory issues.

Nursing knowledge
As the popularity of PDG rises, it is essential for nurses to have a vast knowledge in order to educate, advocate and care for their patients (Lea, 2009). Expanding nursing knowledge about embryonic procedures, educating parents on possible fetal outcomes, and offering guidance through their medical course will help patients make informed choices about their family’s medical future. Families must be aware that these innovative procedures not only impact physical characteristics that are desired, but may alter DNA that could potentially prevent or cause genetic diseases later on in life. It is important that nurses are up to date with risks and benefits of these procedures and how they impact the quality of life of the patient and unborn child. Without knowledge of this new technology and possible outcomes, the patient becomes at risk for making an uneducated decision.

**Access to care**

Although there are many fertility centers that provide IVF, according to Infertility Resources, there are currently 27 locations in the United States with the capability to offer PGD, many of which are located in California (The Fertility Institute, 2013). Centers like The Fertility Institute offer PGD to couples who hope to use tradition IVF methods to conceive a baby. In addition, homosexual couples and those with reproductive health problems, also have the option to use in house egg donors, sperm donors, and surrogates (The Fertility Institute, 2013). Although PGD locations are limited and costly, centers like The Fertility Institute do not discriminate and offer multiple resources to provide successful outcomes.

**Patient centered care**

In a society that demands perfection, there are an increasing number of couples who have turned to science to help create their perfect infant. Having the ability to choose eye color, cholesterol make up, and genetic composition are some of the key points of patient centered care
in the world of “designer babies”. PGD primarily focused on preventative genetic screening in order to decrease the risk of genetic mutations that cause disease. This technology identifies genetic changes in embryos that are created externally via reproductive technology (Lea, 2009). Few areas of focus with PGD include reducing the risk of autosomal recessive inheritance or X-link inheritance. For example, if a couple is trying to conceive and recognize the risk of hemophilia, they would have the opportunity to delete this gene sequence all together or opt for a female embryo. Other illnesses that may be avoided with PGD include cystic fibrosis, sickle cell anemia, and Duchenne muscular dystrophy (Lea, 2009). Once the fertilized embryo is free of mutation, implantation occurs within the uterus of the mother. The use of PGD can identify a newborns risk for disease, help parents learn their risk of having a child with illness, or prevent an illness from affecting a family. Allowing the parents the right to choose the destiny of their unborn child is a direct reflection of patient centered care.

**Patient safety**

PGD is a complex process composed of various invasive procedures including IVF, cell biopsy, genetic analysis and embryo transfer (Hershberger, Schoenfeld & Tur-Kaspa, 2011). Many questions exist regarding the safety of PGD for both the patient and unborn child. Women undergoing PGD are at a high risk for experiencing ovarian hyper-stimulation syndrome, early pregnancy loss, ectopic pregnancies, miscarriages, and procedural complications, such as bleeding, infection, or damage to other internal structures (Mayo Clinic, 2013). During the biopsy, genetic analysis, and transfer phases, the embryos are physically manipulated which may affect embryonic development, rate of implantation, and pregnancy outcome (Turk-Kaspa, 2012). If these procedures are successful, these women are placed at higher obstetrical risk due to implantation of multiple embryos. Research also suggests that these techniques increase the
risk of premature delivery and low birth weight infants (Mayo Clinic, 2013). Complicating matters even further, the long-term outcomes of children conceived using PGD technology remains uncertain, as studies examining these issues are still ongoing and results are not yet available (Hershberger & Pierce, 2010).

**Quality of care**

Quality of care is often associated with safety. Quality care has multiple components; it is safe, effective, patient centered, timely, efficient, and equitable (Mitchell, 2008). Pre-Implantation Genetic Diagnosis and Screening is patient centered. Its purpose is to provide patients with their desired outcome: an infant of a particular gender, free of chromosomal defect, and/or potentially a specific genetic trait. This type of trend is both effective and efficient; The Fertility Institute reports 99.99% accuracy in gender selection and a 92-99% in genetic disease detection (“Sex selecting, 2001-2013”). This trend can be considered both equitable and not. The patient may choose what to screen for and potentially which embryos to implant. However, this service although available to anyone is not affordable to everyone.

**Cost**

PGD is a costly elective procedure offered in specialized clinics. The Fertility Institute offers a wide range of fertility assistance at a substantial price. Before any service is provided, the company requires full compensation. According to the patient’s insurance provider, fertility issues may or may not be covered for some procedures. Many clinics conveniently offer loan payment programs in order to assist with the financial burden. All rates provided are approximated and are quoted if no other health complication exists or arises during the procedures. PGD is offered to patients at a ball park fee of around 21 thousand dollars. This fee includes in vitro fertilization fees, but excludes additional drug prescriptions. These rates may
vary according to location. Additionally, the company provides a disclaimer in which rates are subject to change without prior notice.

**Ease of use**

As expected, PGD is more complicated and time consuming than traditional methods of conception. When compared to complexity of PGD versus IVF only, the ease of use for the families of PGD is very similar. Many IVF centers recommend some genetic counseling and testing; however, with PGD it will likely be more extensive and invasive. With the requirement of ovarian stimulation, the mother may receive hormone injections, ultrasounds, and serial lab tests to monitor the development of follicles. The removal of eggs is typically performed in an office, transvaginally, and under anesthesia. After fertilization, embryos can be biopsied for PGD and later implanted into the mother if desired.

**Interoperability**

While the popularity and usage of PGD has grown significantly over the past few years, the actual success rate is solely dependent on the technology established by IVF. Utilizing the resources of IVF and integrating them to work for PGD is an example of interoperability. PGD requires in vitro fertilization to allow for retrieval of developing embryos (Hershberger, 2010). Once the embryos have been accessed, a second injection is performed to allow for a clear sample of the genetic analysis. From this point forward, the use of PGD technology can focus on the parent’s choice of gender selection or phenotypic genes. Without the usage of IVF, PGD cannot succeed.

**Availability**

Although IVF is provided by 44 states in the U.S. in a variety of clinics and facilities not all provide PGD. In the United States PGD is offered in 14 states, within these states there are
currently 27 centers that provide this service (Preimplantation genetic diagnosis, 2013). For those patients interested in the technology of PGD many must travel out of state in order to receive care.

**Education/training needs of staff or patients**

Ensuring that nurses and other providers working with PGD develop a strong foundation of basic genetic principles will ensure the provision of competent, personalized genomic healthcare (Lea, 2009). Providers must have knowledge about associated concepts including the identification of single gene disorders, chromosomal rearrangements, adult onset genetic disorders, and embryonic matching of human leukocyte antigen (HLA) for sibling stem cell transplantation. They will also require education on the techniques and equipment used during the complex, multistage process of PGD including: IVF, cell biopsy, genetic analysis, and embryo transfer (Hershberger et al., 2011).

Couples undergoing PGD will require extensive patient education understand the potential risks and make informed choices. Hershberger, Schoenfeld & Tur-Kaspa (2011) view nurses and advanced practice nurses as frontline providers of accurate information and education to couples. The benefits and limitations of PGD should be included. The benefits must address the topics of: maximizing the potential for a genetically healthy child, avoiding the anguish of deciding to terminate an abnormal pregnancy, reducing the potential of childhood hospitalizations and decreasing pain and suffering. The discussion of limitations must include: IVF as a prerequisite to PGD, lower pregnancy success rates for IVF with PGD than for IVF alone, the possibility of misdiagnosis, technical difficulties that may be encountered, and the significant financial cost. Additionally, nurses must ensure that couples understand the medical terminology as applied to the process. Information on alternative reproductive options such as
adoption, gamete donation, or traditional prenatal diagnosis should also be included in the education of high-genetic-risk couples (Hershberger et al., 2011).

**Potential ethical issues**

As this innovative technology becomes more prominent in its usage, ethical concerns about its processes arise. “Trait selection and enhancement in embryos raises moral issues involving both individuals and society” (Simmons, 2008). Four chief concerns exist in regards to PGD: safety of embryos, ethical consent, selective accessibility and arguably the most important, playing the role of a creator.

As the PGD technology gains popularity, we must be concerned with the safety of technique while handling these newly created embryos. While a desirable embryo is created and implanted, what happens to those embryos that are not so desirable? Are these embryos stored for further usage, used for experimentation or simply disposed of? Furthermore, does the idea of life begin with implantation or with the unity of a sperm and egg to form a zygote? The PDG process allows parents to select and alter physical characteristics such as eye/hair color, skin complexion and gender of the baby. The manipulation of these traits allows the parents to essentially design their own babies. In altering these physical characteristics, scientist may be inadvertently altering other traits such as personality, abstract thinking, reasoning or pain sensors. These alterations could have a negative impact on the future child. Little research has been conducted to reassure parents that multiple genes are not influenced. Moreover, as the child is concerned, there is no ethical consent as to what genes will be manipulated. This may cause a future disruption in the child’s social development.

Another concern with PDG is the selective accessibility related to cost. Due to the extraordinary costs of the procedure, a limited clientele will be able to utilize this technology.
This population may choose those characteristics for their babies that are seen as desirable within our society. The availability of PDG to a small population “may create a genetic aristocracy and lead to new forms of inequality” (Simmons, 2008).

Finally, one of the most obvious concerns with PGD is the issue of playing the role of a creator. Families must battle this ethical issue as it pertains to their lives spiritually and religiously. One question they must ponder is that of creation and how it is interpreted in their lifestyle and if it is advanced through scientific experimentation.

**Privacy issues**

The Fertility Institute, a leader in PGD, requires an extensive medical history from the mother and the father before prior to their first appointment. Candidates for PGD meet with a geneticists or genetic counselor to discuss their medical history and evaluate their current diseases or future risks of disease to more precisely guide the genetic testing of the embryos. If there are any present diseases, genetics testing will be performed on the parents to confirm diagnoses (The Fertility Institute, 2013). This information is protected by HIPPA laws, however it may be documented in the family’s medical record, and is likely to be passed between doctors, technicians, counselors, and possibly labor and delivery personnel. Any time there is a release or transfer of information there is a risk of a breach in privacy.

**Regulatory issues**

Although the use of PGD is controversial and identifies many ethical issues, currently the United States has not defined any governmental regulation over its use. Directing the care of PGD would involve government interference on issues of personal choices in life or death. While the government continues to battle the issues of abortion and women’s rights, the use of PGD has been left up to the judgment of the providing health care specialist. Within clinics that offer
PGD, some practitioners may choose to screen for only genetic conditions, while others may allow the future parents to select the infant’s gender (The Fertility Institute, 2013). Although the government has not enacted any laws that dictate the usage of PGD, the state of New York has defined standards for their laboratories when collecting specimens for PGD (Baruch, 2008). Laboratories must apply for a permit before submitting an application for approval on their methods of withdrawing samples via IVF (Barauch, 2008). The laboratory methods are then analyzed by the state’s Department of Health’s Clinical Laboratory Evaluation Program (Barauch, 2008). All laboratories that accept cells for PGD usage must have prior approval from the state of New York before proceeding with PGD. For now, regulation of PGD within the United States has been left up to the clinics, although future standardization may be in the near future.

Conclusion
References


