Carrier Disease Screening and Single Gene Testing Informed Consent

Purpose:

Conceptions offers a comprehensive genetic testing program to optimize our patient’s chances of conceiving a healthy child. This program consists of parental genetic carrier screening for single gene mutations and then, if indicated and desired, genetic testing of embryos produced in an in vitro fertilization (IVF) cycle.

Carrier Screening:

Genetic disorders resulting from alterations (also known as mutations) of a single gene account for 10% of pediatric deaths. Unfortunately, in approximately 80% of these cases, there is no family history of genetic disease. Parental genetic screening for these diseases can identify and potentially prevent these inherited conditions. If either or both parental genetic screening tests result in an abnormality, an increased risk of child(ren) with this abnormality may exist. Positive results should be discussed with a physician and a certified genetic counselor. Consideration of IVF, which allows for the genetic testing of your embryo(s) for the condition(s) of concern prior to fertility treatment, may be warranted.

Conceptions utilizes Counsyl as a reference laboratory to identify patients that may be a carrier for a variety of diseases. Based on the couple’s ethnicity, Counsyl can devise reproductive risk ratios when each couple’s test results are paired and analyzed together. This paired analysis provides the most accurate results. While carrier screening testing can be waived, Conceptions strongly recommends that all patients take advantage of this technology as a tool for optimizing your chances for a successful IVF cycle and a healthy baby.

Conceptions recommends the ACOG (American College of Obstetricians and Gynecologists) + ACMG (American College of Medical Geneticists) panel plus MCADD (the most common cause of Sudden Infant Death Syndrome) and PKU (phenylketonuria). Although this is not exhaustive, these recommendations encompass common autosomal recessive single gene disorders in our multicultural, multi-ethnic society.

Pre-Implantation Genetic Diagnosis (PGD) – Single Gene Testing:

For patients with diseases caused by single gene disorders, Conceptions can analyze cells from each embryo that may be affected by a genetic disease known to be present in the family.

CONCEPTIONS REPRODUCTIVE ASSOCIATES OF COLORADO

www.conceptionsrepro.com

INITIAL _______/ ________
Indications:

Couples who are carriers for a specific genetic disorder are at risk for transmitting the disorder to their child. Some examples of these disorders are Cystic Fibrosis, Tay Sachs, Fragile X, or Huntington's disease. The genetic material removed from the embryo at the time of biopsy is limited, so prior knowledge of the genetic mutation carried by the genetic parents and family members may be necessary for the testing to be performed.

PGD can also detect translocations, inversions or other structural chromosomal rearrangements. This is done using specific genetic “probes” selected for individual patients based on their unique abnormality.

Risks and Limitations:

Genetic testing of the embryo has risks and limitations that need to be considered. There is a chance of misdiagnosis due to test error or mosaicism. Mosaicism is when there is more than one chromosomally distinct cell line in the same embryo. This occurs by chance during embryo development and can cause a misdiagnosis if the cells tested are not representative of the entire embryo. This can result in a false negative or a false positive result for the embryo being tested. For this reason, we recommend prenatal testing and screening, as recommended by the American College of Obstetricians and Gynecologists, be strictly followed.

Other possibilities associated with genetic testing of each embryo include no normal embryos for transfer or lack of test results. There is a chance that all embryos that are tested may come back abnormal and therefore, unsuitable for embryo transfer. In these cases, an embryo transfer will not be performed. There is also a chance during preparation, transportation, or analysis of the cells that a result cannot be obtained for a particular embryo or for all embryos.

Not all chromosomal or genetic abnormalities are identifiable and not every gene is analyzed. Therefore, genetic testing of the embryo does not guarantee the birth of a chromosomally and/or genetically normal, healthy child.

Possible Benefits of Single Gene Testing of Your Embryo(s):

Genetic diagnosis of your embryos may increase the possibility of becoming pregnant with a healthy child. Genetic testing of your embryos may improve our ability to recognize possible abnormalities in embryos before placement into the uterus, which may increase implantation (pregnancy) rates. This may also help to reduce the miscarriage rate and reduce the chance of having a child with abnormalities. Information obtained from this testing may also become beneficial for pregnancy attempts in the future.
Alternatives to Genetic Testing of Your Embryo(s):

Standard prenatal testing once you are pregnant, such as chorionic villus sampling (CVS), amniocentesis, blood tests, and/or ultrasound are alternatives to this testing. These options should be discussed thoroughly with your obstetrician or with the person who would be performing or ordering the tests. You may also discuss your options with a genetic counselor. Genetic testing of your embryo(s) is not a substitute for routine prenatal testing. You should undergo recommended prenatal testing based on your age and medical history, even if genetic testing of your embryo(s) has been performed. Testing of your embryos is not required and you should be aware that the procedure is elective.

Consent for Genetic Testing of Your Embryo(s):

We have read the entire consent form or it has been read to us. We have been informed that one or both of us is a carrier for one or more single gene disorder(s). We understand that genetic testing of our embryo(s) has benefits and risks, some of which may be unknown at this time. We understand that this testing cannot detect all abnormalities and that it does not eliminate the potential need for prenatal testing such as chorionic villus sampling or amniocentesis. If pregnancy is achieved, we understand it should be followed by recommended antenatal testing. We understand if we have questions about prenatal testing, we may ask our obstetrician or a genetic counselor. We have been given the opportunity to ask questions about PGD and the contents of this consent form. If we have any additional questions, we may contact our physician. The following initials indicate our position with respect to testing our embryo(s) for a single gene abnormality:

________ We ACCEPT genetic screening of our embryo(s) to rule out a single gene abnormality.

________ We DECLINE genetic screening of our embryo(s) to rule out a single gene abnormality.

Mark R. Bush, M.D., FACOG, FACS
Michael S. Swanson, M.D., FACOG
Dana R. Ambler, D.O., FACOOG
Ryan M. Riggs, M.D., FACOG
Consent to Discard Abnormal Embryo(s):

We understand that by doing genetic testing, some or all of our embryos may come back genetically abnormal and cannot be used for an embryo transfer. This includes any embryo that has aneuploidy or a single gene defect. We hereby give Conceptions permission to discard any genetically abnormal embryo on our behalf.

INITIAL REQUIRED ______/_______

__________________________  ____________________________  
Printed Name of Patient  Printed Name of Partner

__________________________  ____________________________
Signature of Patient  Date  Signature of Partner  Date

Subscribed and sworn to before me this ______day of ________, 20___

My Commission expires: 

__________________________
Notary Public

__________________________
Address