IDENTIFYSGD

Your guide to single gene disorder testing

THE **FOUNDATION** FOR
EMBRYONIC COMPETENCE

ADVANCING KNOWLEDGE. ENHANCING OUTCOMES.

"The tallest oak in the forest was once just a little nut that held its ground."

– Unknown –

GROWING FAMILIES

What is Identify**SGD**?

IdentifySGD, available only through the Foundation for Embryonic Competence (FEC), is a unique test that is designed to detect genetic alterations associated with a specific single gene disorder in an embryo prior to transfer during an in vitro fertilization (IVF) cycle.

A single gene disorder is a condition that is caused by a known alteration or mutation in one of more than 20,000 genes found in nearly every cell of the human body.

The condition may be inherited through one or more generations in a family, or inherited from both members of a couple who are carriers of the same condition. Examples of single gene disorders are fragile X syndrome, Tay-Sachs disease, sickle-cell anemia, spinal muscular atrophy, Huntington's disease, Duchenne muscular dystrophy, and cystic fibrosis.

IdentifySGD is one kind of preimplantation genetic diagnosis (genetic testing before embryo transfer) that can be of benefit to an individual or couple at increased risk of having a child with a single gene disorder based on family or medical history.

Which single gene disorders can be identified through testing?

The FEC has successfully tested for a variety of genetic disorders and can test for nearly all single gene disorders, with some rare exceptions. There are also other types of preimplantation genetic diagnosis that look for extra or missing chromosomes (eg, Down syndrome). Talk to your doctor, nurse, or genetic counselor about specific single gene disorders for which you may consider testing.

How does Identify**SGD** work?

To initiate IdentifySGD testing, you must first complete a phase of preparation called work-up, or validation (*see Fig. A*). During this phase, we will require blood work from you and your partner. Depending on the disorder(s) for which you are testing, we will likely ask that other family members submit a buccal (cheek) swab, if possible, to assist with this process. Using these samples, the FEC will customize a unique test for your family.

With findings from a unique test created for each family, providers and patients can make better decisions about which embryos should be considered for IVF.

Once validation is complete, the lab is ready to accept genetic material from the embryo during the next phase called the biopsy phase (*see Fig. B*). IdentifySGD requires a sample of approximately 5 to 10 cells biopsied (safely removed) from an embryo. These cells are removed from a specific area of the embryo called the trophectoderm at a specific time in embryonic development. Trophectoderm biopsies have been proven not to harm the embryos.

Using the unique test developed for your family, our staff will determine if each embryo is positive or negative for the variation or mutation of interest. All results are reviewed by our laboratory director before the results are reported.

Additionally, all embryos tested with IdentifySGD will receive SelectCCS, a proprietary test that screens embryos for too many or too few chromosomes, a condition known as aneuploidy.

By the end of the IdentifySGD process, providers and patients can make better decisions about which embryos should be considered for IVF transfer based on their genetic makeup.

Fig. A

VALIDATION PHASE

STEP 1
Review genetic reports

STEP 2
Collect samples

STEP 3
Develop custom test

STEP 4
Notify IVF team of phase completion

Fig. B

BIOPSY PHASE

STEP 1
Receive embryo biopsy

STEP 2
Analyze DNA

STEP 3
Issue reports

STEP 4
Schedule IVF transfer

Explore other tests at FEClabs.org

How is Identify**SGD** different from older or other forms of single gene disorder testing?

The FEC creates a unique test for each patient to target the specific gene alteration(s) or mutation(s) of interest whenever possible. Many other laboratories are not able to do this. Additionally, our technology allows us to select markers as close as possible to the mutation site to reduce the risk of recombination (rearrangement of genetic material) and provide the most accurate results clinically possible.

Can you test for more than one disorder on a single biopsy?

Yes, to date the FEC has tested up to 3 different disorders using a single biopsy.

"Thank you for helping my cousin realize her dream of parenthood and for helping us stop the line of this disease through our family."

—A family member of an IdentifySGD patient

What happens to my embryos during the Identify**SGD** process?

Your embryos never leave the care and security of your physician's IVF laboratory. Embryos are safely cryopreserved while awaiting the IdentifySGD test results and subsequently transferred in a frozen embryo transfer (FET) cycle.

How accurate is Identify**SGD**?

IdentifySGD has been demonstrated to be at least 98% accurate in screening for single gene disorders; however, like any test, false positives and negatives can occur. When a pregnancy occurs after the transfer of embryos analyzed by IdentifySGD, it is recommended that results are confirmed by chorionic villus sampling (CVS) or amniocentesis. These tests can be coordinated through your obstetrician's office.

What are the limitations of Identify**SGD**?

Not all genetic defects can be detected using IdentifySGD. Also, when undergoing this testing, there is the risk of inaccurate results. IdentifySGD offers no guarantee that pregnancy will occur or that a healthy child (completely free from birth defects or genetic defects) will result.

How long before results are available?

The IdentifySGD validation phase, during which a customized test for the gene mutation of interest is developed for your family, takes approximately 4 to 8 weeks. Once you start cycling and the FEC receives embryo biopsies, the IdentifySGD results will be ready in about 2 weeks. Your physician will receive an in-depth report that will indicate which embryos are positive for the genetic variation or mutation of interest.

What does Identify**SGD** cost?

Speak with your IVF team about the costs and benefits of IdentifySGD and to determine your insurance coverage, if any.

Speak with your doctor or genetic counselor to determine if Identify**SGD** is right for you.

ABOUT THE FEC

The Foundation for Embryonic Competence (FEC) is a nonprofit organization dedicated to advancing knowledge and enhancing outcomes in embryonic research, diagnosis, and education. SelectCCS and IdentifySGD are owned and operated by the FEC. All proceeds from SelectCCS and IdentifySGD are used to support research and education.

See our story at FEClabs.org

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140 Allen Road, Suite 300, Basking Ridge, NJ 07920
908.580.1200 • contact@FEClabs.org