INFORMATION SHEET

Genetic Carrier Screening

This information sheet is written for all individuals and couples planning fertility treatment, to provide an overview on genetic conditions and genetic screening options. It is relevant to individuals and couples using their own eggs and sperm, or those using donor eggs, sperm, or embryos. This is a general information sheet, and it is intended as a guide. We understand that everyone's circumstances are unique and one of our Genetic Counsellors can assist in exploring your specific situation if needed.

Genetic Conditions

We are all carriers for genetic conditions.
Every reproductive couple have a chance of having a child affected by a genetic condition. Most children with genetic conditions are born to reproductive couples who have no known family history of the condition.

Recessive genetic conditions These occur when both biological parents are carriers of the same recessive genetic condition. Carriers have one normal copy and one faulty copy of the recessive gene and are usually healthy. If both biological parents pass
a faulty copy of the recessive gene on to offspring, they will be affected by the condition. Some of the most common recessive genetic conditions are Thalassaemia, Cystic Fibrosis and Spinal Muscular Atrophy, but there are many more.

X-linked genetic conditions These occur when a female biological parent is a carrier for an X-linked genetic condition. Carrier females have one normal copy and one faulty copy of the X-linked gene and may or may not have symptoms of the condition. If the female passes a faulty copy of the X-linked gene on to male offspring, they will be affected by the condition. If the female passes the faulty copy of the X-linked gene on to female offspring, they may be affected by the condition. One of the most common X-linked genetic conditions is Fragile X syndrome, but there are many more.

Genetic Carrier Screening (GCS)

Genetic Carrier Screening (GCS) is a type of genetic testing used to determine if a reproductive couple have a high risk or low risk of having a child affected by a genetic condition. GCS can be performed for a varying number of genetic conditions. Limited GCS will test for some of the most common genetic conditions or expanded GCS will test for many genetic conditions. It is not possible to screen for all possible genetic conditions (e.g 200 to 1000 conditions).

Some GCS tests are individual tests which have individual reports, and the conditions carried by individual are identified. Some GCS tests are couple-based tests which have a couple report only, and the conditions carried by each individual are NOT identified (unless both reproductive partners are a carriers for the same condition).

Recommendation for GCS

It is recommended that all individuals and couples consider GCS before planning to conceive, with or without Assisted Reproductive Treatment (ART). An egg provider is usually screened for recessive and X-linked genetic conditions. A sperm provider is usually screened for recessive genetic conditions only.

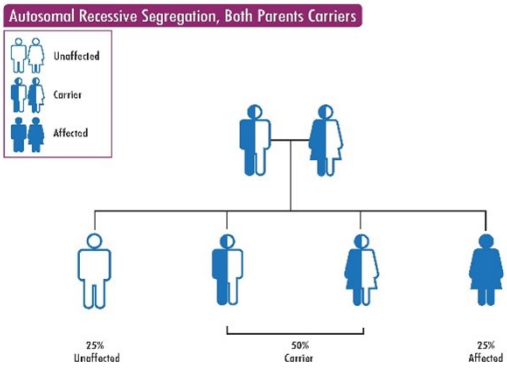
Individuals and couples using their own eggs and sperm, or known donor eggs, sperm or embryos, can choose to have limited or expanded GCS, an individual test or a couple-based test, or may decide not to proceed with any GCS. If testing one reproductive partner at a time, using an individual test, screening should start with the egg provider, to screen for recessive and X-linked conditions. Alternatively, both reproductive partners can be screened simultaneously via either an individual test, or couple-based test.

Individuals and couples using identity release (IR) donor eggs, sperm, or embryos, will find that limited or expanded GCS has already been performed in their donors. The type of screening and level of screening performed will depend on when they were recruited by the clinic, and whether they are local or international donors. All newly recruited IR donors will have expanded screening for many genetic conditions.

Reproductive Genetic Carrier Screening

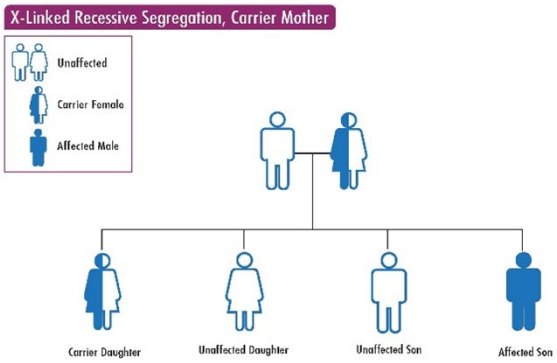
What is the risk to offspring if both reproductive partners are carriers for the same recessive genetic condition?

When both reproductive partners are carriers for the same recessive genetic condition, there is a 1 in 4 (25%) risk of having a child affected by the condition (see below). If you are using your own eggs and sperm, or a known donor, there may be options available to test the embryos for the genetic condition to reduce the chance of having an affected child. If you are using an IR donor, then you will be required to select another donor.



What is the risk to offspring if an egg provider is a carrier for an X-linked genetic condition?

If an egg provider is a carrier for an X-linked genetic condition, there is a 1 in 2 risk (50%) this will be passed on to offspring (see below). If passed on to a male offspring, they will be affected by the condition. If passed on to a female offspring, they may be affected by the condition, or be a healthy carrier for the condition. If you are using your own eggs and sperm, or a known donor, there may be options available to test the embryos for the genetic condition to reduce the chance of having an affected child.



What is the risk to offspring if one reproductive partner is a carrier for a recessive genetic condition?

If one reproductive partner is a carrier for a recessive genetic condition, and the other reproductive partner is at low risk of being a carrier for the same condition, there is a low risk of having a child affected by the condition. When an individual screens low risk for a condition, there remains a residual risk they may still be a carrier for the condition. Therefore, the risk of having a child affected by a condition is never reduced to zero.

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Reproductive Genetic Carrier Screening

If I am using an egg or sperm donor, how do I arrange GCS?

Once you have selected a donor, a genetic counsellor will contact you for an appointment to discuss GCS. If you are using an IR donor, the genetic counsellor will inform you of the type of screening that has been performed in your selected donor and the type of screening that you are required to have to proceed with treatment using that donor. If you are using a known donor, the genetic counsellor will inform you about GCS options so you can decide what you would like to do. If you are using a known donor, GCS is optional. However, if you decline the minimum recommended level of screening (Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X syndrome) you will be required to sign a consent form to acknowledge your choice.

I already had GCS previously. How do I select an IR donor?

After you have had an initial counselling appointment, you will be given access to the donor portal. If you have already had GCS previously, please advise the donor and/or genetics teams. The genetics team will review your GCS results and provide advice regarding donor selection. If you are a known carrier for a recessive genetic condition, you are required to select an IR donor who has screened low risk for that condition. It is possible that you may be required to have additional GCS to use a particular donor. If you decline additional GCS, then you may be limited in the donors you can select from.

If I am using donor embryos, how do I learn about GCS?

Once you have selected donor embryos, a genetic counsellor will contact you for an appointment to discuss GCS. For IR donor embryos, the egg and sperm providers have been screened for many recessive and X-linked genetic conditions. There will be a low risk of having a child affected by the recessive and X-linked conditions tested for. When an individual screens low risk for a condition, there remains a residual risk they may still be a carrier for the condition. Therefore, the risk of having a child affected by a condition is never reduced to zero. If you are using known donor embryos, the genetic counsellor will inform you about GCS options so you can decide what you would like to do. If you are using known donor embryos, GCS is optional. However, if you decline the minimum recommended level of screening (Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X syndrome) you will be required to sign a consent form to acknowledge your choice.

Please call 8080 8933 or email genetics@newlifeivf.com.au to speak with a member of our Genetics Team and obtain additional information about GCS or your unique situation.