

Health and genetic screening considerations when using a known gamete (sperm or egg) donor



When choosing an egg or sperm donor, there are many practical and psychological factors to consider. One consideration should be the health and family history of your chosen donor.

Genea encourages recipients of known donors to obtain general personal and family health history information about themselves and their chosen donor to help assess if there might be an increased chance of having a child with an inherited health issue. Genetic carrier screening can also be considered.

Suggested health history questions to ask your family and donor about:

- Genetic conditions such as cystic fibrosis, spinal muscular atrophy, fragile X syndrome, thalassemia
- Pregnancy issues (fertility issues, pregnancy loss or stillbirth)

- Intellectual disability, developmental delay and congenital health issues (babies or children with a health condition)
- Early onset cancer diagnoses (<50years), or the same type of cancer diagnosis in multiple family members
- Ashkenazi Jewish Ancestry (higher chance of being a carrier of certain genetic conditions for individuals with this ancestry)
- Neurological or muscle conditions
- Significant mental health conditions

Background of genetics

We all have about 20,000 genes, which are the instructions for how our bodies grow and function. We have two copies of each gene, one inherited from the egg and the other inherited from the sperm.

Autosomal dominant inheritance

There are certain conditions which occur when an individual has a variant/alteration on one copy of a gene, causing it not to work properly. This is known as autosomal dominant inheritance (e.g. Huntington disease, BRCA). If your donor has a personal or family history of a dominant condition, there is a 50% chance your child will inherit that condition. If you establish that your known donor has or is suspected of having a dominant genetic condition, we recommend you discuss this with your fertility specialist or a Genea genetic counsellor.

Autosomal recessive inheritance

There is another group of genetic conditions known as recessive conditions. These occur when an individual inherits a variant in both copies of a gene. If an individual has a variant in one copy of a recessive gene only, but the other copy is working, this means the individual is a healthy carrier of that condition. For autosomal recessive genes, carriers generally do not develop symptoms related to the condition. It is common to be a healthy carrier of several recessive genetic conditions.

If both an egg and a sperm provider carry the same genetic condition, any children conceived each have a 25% (or 1 in 4) chance of being affected with that condition (e.g. cystic fibrosis). However, if an egg and a sperm provider do not carry the same condition, then any offspring are most unlikely to have that condition.

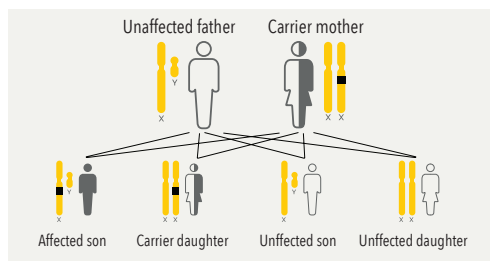


Fig 1. Autosomal recessive inheritance

X-linked inheritance

Females can be healthy carriers of certain conditions and be at risk of having affected males and sometimes females (e.g. fragile X syndrome, which is a common cause of intellectual disability and autism). This is known as X-linked inheritance. Testing of the female donor recipient or egg donor for X-linked conditions could be considered. These results would be relevant for your children regardless of the sperm provider or sperm donor you choose.

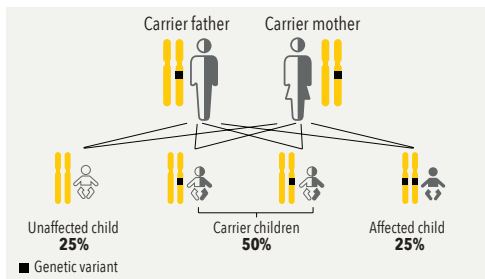


Fig.2 X-linked inheritance

Genetic Carrier Screening

Genetic carrier screening is a blood or saliva test on an egg provider and a sperm provider which can identify if they have an increased chance of conceiving a child affected with certain serious childhood onset, recessive and X-linked genetic conditions. If there is a reported increased chance, there are reproductive options available to you including:

- testing embryos via IVF and Preimplantation Genetic Testing (PGT)
- testing a pregnancy via a CVS or amniocentesis or;
- testing the baby at birth to allow for early treatment.

You may decide to use a different donor.

What does it screen for?

Genetic carrier screening does not test for autosomal dominant conditions.

Genetic carrier screening panels can determine if individuals carry a certain number of X-linked or recessive genetic conditions. Some panels only test for a limited number of common conditions in the population (e.g. cystic fibrosis, spinal muscular atrophy, fragile X syndrome), other panels can screen for over 500 common and rare conditions.

Some tests have Medicare rebates and others have out-of-pocket fees.

If you are interested in reproductive carrier screening, we recommend you contact the Genea Genetic Counsellors to book an appointment to discuss your options.

You may want to start with just testing you, however, testing both you and your donor at the same time will save time as results can take up to 4 weeks to be reported.

To book an appointment,
scan the QR code or email
genetic.counsellors@genea.com.au

