PATIENT INFORMATION

Karyotype

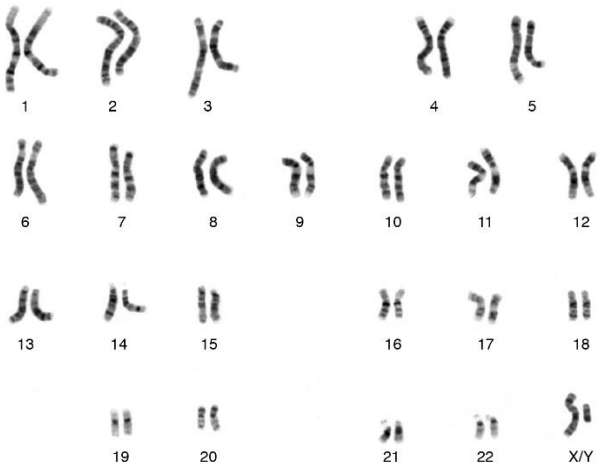
What is a karyotype?

A karyotype is a profile or 'picture' of a person's chromosomes. Karyotyping, also known as chromosome analysis or chromosome studies, examines the size, quantity, and arrangement of our chromosomes. As some chromosome changes are associated with health conditions and fertility challenges, a karyotype can be a useful way of finding out if an individual has a normal quantity and arrangement of chromosome material.

At Nº1 Fertility, a karyotype analysis is part of your initial work up as a patient. Karyotype analysis requires a blood test and is bulk billed to Medicare. This test provides important information about a person's chromosome make up, which helps to facilitate the most appropriate treatment and testing options within your IVF process. As such, karyotyping is a requirement for all patients, partners, and donors at Nº1 Fertility.

What are chromosomes?

Chromosomes are thread-like structures of genetic information that are contained inside within the cells in our body. Most cells in our body contain a full set of 23 pairs of chromosomes, and each chromosome contains genes essential for our growth, development, and healthy function. Genes are made up of DNA, which is packaged into larger structures called chromosomes, that each contain thousands of functional genes. Genes provide specific instructions for our body to grow, develop, and function.



We usually have 46 chromosomes, which come in 23 pairs numbered 1 through 22 by size. The 23rd pair are sex chromosomes that typically determine our biological sex. Usually, two X chromosomes (XX) are found in biological females, and one X and one Y (XY) are found in biological males. We typically inherit one copy of each chromosome from an egg and one from a sperm.

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Why is a karyotype important?

Karyotype analysis can identify chromosome changes which increase an individual or couple's reproductive risk i.e. increase the chance of conceiving a pregnancy with genetic health conditions. Certain chromosome changes can also be associated with challenges in achieving a pregnancy in the first instance. An example of this is individuals who carry structural rearrangements in their chromosomes. Structural rearrangements are changes to the normal size and/or arrangement of chromosomes that can cause subfertility. In IVF, we typically see healthy individuals who carry balanced chromosome structural rearrangements. Many of these individuals not aware they carry a structural rearrangement until they have trouble trying to conceive. If a parent or donor carries a structural rearrangement, it is possible for created embryos to inherit a balanced or unbalanced quantity of chromosomes. Each embryo may have either a normal, balanced quantity of chromosomes; an unbalanced quantity of chromosomes; or a balanced rearrangement of chromosomes like the parent or donor with the structural rearrangement (please see *Patient Information: PGT-SR* for further information).

Additionally, some individuals carry extra or missing 'sex chromosomes' that can explain their difficulties conceiving. Examples include Turner Syndrome (XO), Klinefelter Syndrome (XXY), and Jacob Syndrome (XYY).

It is important to note that individuals with chromosome changes can have healthy children, however often they have reproductive challenges including recurrent miscarriage or difficulties falling pregnant.

What if my karyotype is 'abnormal'?

If a patient, partner, or donor is found to carry a chromosome change or rearrangement that increases their reproductive risk, there might be options for targeted forms of preimplantation testing through IVF, for example, Preimplantation Genetic Testing for a Structural Arrangement (please see *Patient Information: PGT-SR* for further information).

How will I find out if my karyotype is abnormal?

If your karyotype report identifies a chromosome rearrangement or an abnormality, one of our doctors or genetic counsellors will call you to arrange a consultation to help you understand your result and discuss any genetic testing options available to you.

How long do karyotype test results take?

Karyotype results typically take between 8-10 weeks to be reported from the lab.

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Nº1 FERTILITY

Can I start IVF treatment if my karyotype is not back yet?

In best practice, karyotype results are reported prior to beginning IVF treatment. However, we understand time is precious. If you have had the opportunity to understand the implications of an 'abnormal' karyotype finding and wish to commence treatment prior to receiving your results, we will support you to do so.

Questions?

If you have read this information and have questions, please contact the Nº1 Genetics Department on 9132 9600 or email genetics@number1fertility.com to organise a time to speak to one of our Genetic Counsellors.

The information provided above is intended for educational purposes only and should not be used as a substitute or replacement for medical advice received from a medical professional. It is important to discuss your individual circumstances and situation with your treating doctor.

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