Gloucestershire Hospitals
NHS Foundation Trust

Patient Information

Karyotyping and cystic fibrosis screening

Introduction

This leaflet has been given to you to answer some of the questions you may have about the karyotyping and cystic fibrosis screening which has been recommended for you.

There are many reasons why a man might have a very low sperm count, or no sperm at all (azoospermia). The cause is sometimes genetic, which means that a subfertile man might pass the same type of sub fertility on to his son.

New techniques such as Intra cytoplasmic sperm injection (ICSI) can help a couple to achieve a pregnancy despite a very low count or azoospermia, but there is understandable concern about the possibility of passing a genetic problem onto children. Leaflet GHPI0466 Intracytoplasmic Sperm Injection (ICSI) is available for more information.

This leaflet explains why, if a sperm count is below a certain level, we recommend further blood tests before In Vitro Fertilization (IVF) or ICSI.

The results of the test may help couples decide whether the level of risk to their future children is acceptable or not.

We deliberately say ‘can help’ because the tests are not perfect. In other words, no tests can guarantee a perfect outcome.

Recommended tests

Karyotyping (analysis of chromosomes)

Chromosomes carry the genetic information (DNA) that makes all human beings unique. Rarely, problems with sperm production arise because a man is born without a full set of chromosomes. Karyotyping involves giving a small blood sample. The analysis of the sample then involves counting the number of chromosomes present. The result is usually available after 8 weeks.

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Y chromosome Microdeletions (missing segments)

The male chromosomes include X and Y. If the test shows missing segments on the Y chromosome, the doctor may offer you treatment for retrieving sperm called surgical sperm recovery. Please refer to leaflet GHPI0455 Severe oligospermia and azoospermia.

Cystic Fibrosis Screening

Up to 5 to 10% of azoospermic (absence of sperm) men may have a condition called congenital bilateral absence of vas deferens (CBAVD). This means that neither vas deferens (the tubes that carry sperm from the testes) is present. The condition is associated with cystic fibrosis which is a disease that affects the lung's capacity to cough up phlegm. Screening involves giving a small blood sample to see if you carry one of the genes that causes cystic fibrosis.

Contact information

If you have any questions, please feel free to contact the fertility nurses through Mrs K Reddy's secretary on the number below.

Cotswold Fertility Unit
Tel: 0300 422 3128
Monday to Friday, 8:00am to 4:00pm
Website: www.cotswoldfertilityunit.co.uk

Further information

Human Fertilisation Embryology Authority
Website: www.hfea.gov.uk/

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