Rodinia®

infertility test

Personalized

insights for **family** planning

MEDICOVER GENETICS

WHAT IS **Rodinia**?

Rodinia is a genetic test for **infertility** that screens for genetic variants (mutations) in individuals who have difficulty achieving pregnancy. Rodinia can guide towards the optimal clinical management plan for each couple or individual wishing to have a child by identifying disorders of sex chromosomes and genetic variants associated with infertility.

CLINICAL UTILITY

Infertility is considered a major disorder of the reproductive system, and it **affects approximately 1 in 6 couples worldwide**[1].

Biochemical and instrumental testing, such as hormone checks, ultrasounds and semen analysis, can identify up to 65% of infertility cases[2]. If the cause of infertility remains undetermined, genetic testing can be offered, as a significant number of infertility cases are attributed to **genetic mutations**. In fact, up to 10-15% of infertile individuals have a genetic mutation linked with infertility[3]. Genetic testing can provide deep insight that acts as a **valuable prognostic tool**. Rodinia can guide you to take **informed** and **accurate decisions** on the **best clinical management** and **treatment** for your patient, helping to increase their chances of achieving a pregnancy.

WHO IS **Rodinia** FOR?

Female and Male Infertility Panels are applicable for:

- couples or individuals experiencing pregnancy delay
- individuals with specific phenotype indicating a genetic syndrome associated with a sex chromosome aneuploidy
- females with irregular or absent menstruation
- males with low sperm count, irregular sperm form, or movement
- any individual or couple that will undergo assisted reproductive technology (ART) treatment
- candidates for sperm or oocyte donation
- individuals with family history of infertility

Thrombophilia and NAIT Panel is applicable for:

- couples or individuals experiencing recurrent pregnancy loss
- individuals with early-age onset or with a strong family history of thrombotic events
- individuals with recurrent episodes of thrombosis
- neonates with thrombocytopenia
- neonates with unexplained thrombosis

1. *Agarwal, Ashok, et al. "A Unique View on Male Infertility Around the Globe." Reproductive Biology and Endocrinology, vol. 13, no. 1, BioMed Central, Apr. 2015*
2. *Cariati, Federica, et al. "The Evolving Role of Genetic Tests in Reproductive Medicine." Journal of Translational Medicine, vol. 17, no. 1, BioMed Central, Aug. 2019*
3. *Dukhovny, Stephanie, and Louise Wilkins-Haug. "Genetic Basis of Female Infertility." Clinical Genomics: Practical Applications in Adult Patient Care, 1e Eds. Michael F. Murray, et al. McGraw Hill, 2014*

WHAT DOES **Rodinia** TEST FOR?

Rodinia **Female** and **Male Infertility panels** screen numerous genetic variants associated with infertility, and test for **whole**, **partial** and **mosaic sex chromosome changes**. Rodinia **Thrombophilia and NAIT Panel** screens for 22 genetic variants in 17 genes.

FEMALE INFERTILITY PANEL

55 genes, X chromosome aneuploidies

Screens for genetic variants associated with female infertility and includes disorders such as:

- primary ovarian insufficiency
- polycystic ovary syndrome
- ovarian hyperstimulation syndrome
- hypogonadotropic hypogonadism disorders like Kallmann syndrome

Early interventions to preserve fertility

Early identification of women with primary ovarian insufficiency can determine the need for oocyte harvesting and cryopreservation at a young age to preserve fertility and enable the patient to have a child in the future.

MALE INFERTILITY PANEL

40 genes, X and Y chromosome aneuploidies, including Y-chromosome microdeletions

Screens for genetic variants that can cause male factor infertility, including:

- sex chromosome changes
- hypogonadotropic hypogonadism disorders like Kallmann syndrome
- azoospermia

Prognostic value for treatment

*Azoospermic men have a higher prevalence of microdeletions on the Y-chromosome than men with normal sperm and **should receive genetic testing to clarify etiology prior to treatment**[4]. Of the 3 AZF deletions that are associated with azoospermia, only one is associated with a chance of recovering sperm through testicular sperm extraction treatment (TESE).*

THROMBOPHILIA AND NAIT PANEL

22 genetic variants

Screens for specific genetic variants associated with recurrent pregnancy loss, thrombophilia and NAIT.

Can be selected as an Add-on Panel to the Female or Male Panel, or as a Stand-alone Panel.

Guidance on treatment decisions

Identifying variants associated with recurrent pregnancy loss can determine the need for receiving treatment, such as hormonal therapy or anticoagulants, which may reduce or prevent complications during pregnancy and delivery.

4. *Pfeifer, Samantha M., et al. "Diagnostic Evaluation of the Infertile Male: A Committee Opinion." Practice Committee of the American Society for Reproductive Medicine. Fertility and Sterility, vol. 103, no. 3, Elsevier BV, Mar. 2015, pp. e18–25*

WHY RECOMMEND **Rodinia** TO MY PATIENTS?

The genetic insight provided by Rodinia on the reproductive health of an individual or couple can:

- guide treatment or clinical management options
- enable early interventions to preserve fertility
- provide accurate prognostic assessment which can lead to a personalized clinical course of action

CASE STUDY

33-year-old man with 2 failed in vitro fertilization (IVF) attempts

Semen analysis shows

- nil sperm concentration
- nil sperm motility

Patient referred for genetic testing as per azoospermia recommendations[4].

After Rodinia Infertility testing results

A **homozygous 5T variant** in the CFTR gene was detected, which is associated with:

- congenital bilateral absence of the vas deferens (CBAVD), a condition that causes **obstructive azoospermia**

As **spermatogenesis is normal**, the patient can proceed with assisted reproductive technology procedures such as IVF and intracytoplasmic sperm injection with the use of retrieved sperm from the testicles, epididymis or vas deferens. Another option would be microsurgery to repair blocked tubes in the reproductive tract.

TECHNOLOGICAL ADVANTAGES

TARGETED TECHNOLOGY

Rodinia is based on a novel, **Target Capture Enrichment Technology** which utilizes Next Generation Sequencing, and has been thoroughly validated for its accuracy and precision.

NOVEL BIOINFORMATICS

Innovative bioinformatic pipelines analyze the sequencing data produced from each sample, increasing the sensitivity and specificity of Rodinia.

FULL COVERAGE

Rodinia screens for **all coding regions*** of the genes tested, detecting:

- single nucleotide variants (SNVs)
- small insertions and deletions (INDELs)
- copy number variants (CNVs)

The Thrombophilia and NAIT Panel targets specific variants.

THOROUGH CHROMOSOMAL ANALYSIS

X and Y chromosome analysis for whole copy number changes, and partial changes down to 10Mb. Mosaicism detection in sex chromosomes down to 15%.

**Exceptions on regions containing repeats, sequences of high homology (pseudogenes and segmental duplications) or extreme GC-content.*

BENEFITS OF **Rodinia**

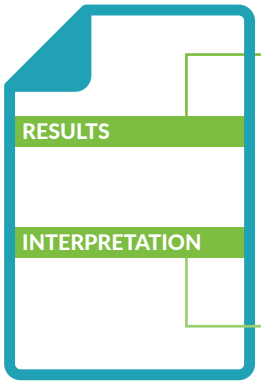
SAFE
Non-invasive sample collection

VALUABLE
Guides patient-specific clinical management

INFORMATIVE
Extensive list of clinically actionable mutations

THOROUGH
Full exonic coverage*

WHAT WILL THE **REPORT** TELL ME?



CLINICALLY SIGNIFICANT VARIANT DETECTED
A pathogenic or likely pathogenic variant has been identified in a gene tested, which is associated with infertility.

NO CLINICALLY SIGNIFICANT VARIANT DETECTED
No disease-causing genetic variant has been identified in the genes tested.

VARIANT OF UNCERTAIN SIGNIFICANCE
A genetic change has been detected, but it is currently unknown whether that change is associated with a genetic disorder.

Summary of the results and recommendations

Carrier status will not be reported for recessive conditions.
The Thrombophilia and NAIT Panel reports the genotype of the variants tested.

WHAT CAN I DO AFTER **Rodinia**?

By identifying the underlying genetic cause of infertility, Rodinia can help you to:

- determine accurate prognosis for your patient
- evaluate and choose the most optimal treatment for your patient
- manage clinical care by being aware of potential complications, like ovarian hyperstimulation
- advise your patient on how to proceed with pregnancy planning, if applicable
- recommend testing to family members, if applicable

**Exceptions apply*

HOW TO ADMINISTER **Rodinia**?

Recommend the appropriate **Rodinia** panel to your patient

Collect a buccal swab from your patient

Send the sample to **Medicover Genetics**

The sample will be analyzed at **Medicover Genetics** laboratories

Results will be sent to you within 2-4 weeks from sample receipt

MORE **QUESTIONS**?

If you have additional questions or concerns, please contact us at **info.genetics@medicover.com**

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