Adventia®
carrier screening

# Genetic insights to empower your life decisions

MEDICOVER GENETICS

# WHAT IS **Adventia**?

Adventia **carrier screening** is a genetic test which determines whether a phenotypically healthy person is a carrier of a genetic disorder. The genetic insight provided by Adventia can inform, guide, and empower people on their **reproductive choices**, and minimize the risk of people who are carriers transmitting a genetic disorder to their children.

## CLINICAL UTILITY

**1 in 4 people** is a **carrier** of a genetic disorder[1]. Genetic disorders may be rare, with varying prevalence, but can have adverse effects on health and development, or even be life-threatening. Without carrier screening, asymptomatic carriers of recessive mutations often discover their carrier status only after they have an affected child.

Early knowledge of one's carrier status and of their subsequent risk of transmitting a genetic disorder to their children can:

- help individuals evaluate their reproductive options to prevent that risk
- result in earlier interventions and therapies where available
- lead to better clinical management of affected children

### RECOMMENDATIONS AND STATEMENTS FROM INTERNATIONAL ORGANIZATIONS:

- Carrier screening information should be provided to all pregnant women[2,3].
- Published evidence supports clinical utility for carrier screening of multiple conditions simultaneously[4].
- Carrier screening panels should be neutral to ethnicity and population, and more inclusive of diverse populations in order to promote equality and inclusion[4].

# WHO IS **Adventia** FOR?

- Couples planning to start their families and wanting to know about their carrier status
- Any individual or couple going through assisted reproduction, including in vitro fertilization (IVF)
- Sperm and oocyte donors, and recipients of sperm or oocyte donation
- Couples who are already pregnant and want to know whether their child has a risk of having a genetic disorder
- High-risk population groups for specific disorders
- People with a family history of a genetic mutation
- Any individual wishing to know more about their genetic profile


1. *Lazarin, Gabriel A., et al. "An Empirical Estimate of Carrier Frequencies for 400+ Causal Mendelian Variants: Results from an Ethnically Diverse Clinical Sample of 23,453 Individuals." Genetics in Medicine, vol. 15, no. 3, 2012, pp. 178–186*
2. *American College of Obstetricians and Gynecologists "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstetrics & Gynecology, vol. 129, no. 3, 2017*
3. *Edwards, Janice G., et al. "Expanded Carrier Screening in Reproductive Medicine—Points to Consider." Obstetrics & Gynecology, vol. 125, no. 3, 2015, pp. 653–662*
4. *Gregg, Anthony R., et al. "Screening for Autosomal Recessive and X-Linked Conditions during Pregnancy and Preconception: A Practice Resource of the American College of Medical Genetics and Genomics (ACMG)." Genetics in Medicine, vol. 23, no. 10, 2021, pp. 1793–1806*

# **Adventia** PANELS

## 1-2 gene(s) FOCUS PANELS

Six panels for **highly frequent** and **severe** genetic disorders:

| PANEL | TESTING METHODOLOGY | DETECTION RATE |
|---|---|---|
| **A-Thalassemia** | MLPA | >90% |
| **B-Haemoglobinopathies**<br>*B-Thalassemia & Sickle Cell Anemia* | Sanger | >95% |
| **Cystic Fibrosis** | NGS | >97% |
| **Dystrophinopathies**<br>*Duchenne Muscular Dystrophy & Becker Muscular Dystrophy* | MLPA | >75% |
| **Fragile X Syndrome** | Fragment analysis (PCR) | >99% |
| **Spinal Muscular Atrophy** | MLPA analysis | >75% |

*MLPA: Multiplex Ligation-dependent Probe Amplification, NGS: Next Generation Sequencing, PCR: Polymerase Chain Reaction*

## 22 genes CORE PANEL

Single panel screening for 22 genes associated with genetic disorders of **high incidence** and **severity**. The Core panel includes all disorders tested in the Focus panels, and other disorders including: *Phenylalanine Hydroxylase Deficiency, Fanconi Anemia Group C* and *Tay-Sachs disease*.

## 231 genes COMPREHENSIVE PANEL

Single panel screening for 231 genes associated with disorders that have **moderate to severe**, **well-defined phenotype** and **high cumulative frequency**. The Comprehensive panel includes all disorders of the Core panel, and covers a wide range of metabolic, cardiovascular, haematological, immunological, neurological and pulmonary disorders, amongst others.

Testing for Core and Comprehensive panels is performed via **Target Capture Enrichment Technology** utilizing **NGS**.
*Exceptions include Spinal Muscular Atrophy and Fragile X syndrome which are performed via the methodologies described in the Focus panels.*
*Testing for A-Thalassemia, B-Haemoglobinopathies and Dystrophinopathies via the Core and Comprehensive panels yields higher detection rates.*
*For a complete list of the disorders tested by Adventia Core and Comprehensive panels please visit www.medicover-genetics.com*

# WHY RECOMMEND **Adventia** CARRIER SCREENING?

Adventia tests for autosomal recessive and X-linked disorders, which:

- have moderate to severe phenotype
- are high in carrier frequency
- can severely compromise quality of life
- may be manageable through early interventions

## CASE STUDY

Couple with one affected child with Cystic Fibrosis (CF)
*Over 2,000 mutations could cause CF*

Prior testing identified:

- a well-known CF-causing mutation in the mother
- no pathogenic CF-causing mutation in the father

### Adventia Core panel testing results

A heterozygous, **single exon deletion** related with CF in the father. This mutation is characterized by its:

- exceptionally small size
- extremely challenging detection due to the mutation type (copy number variant)

The detection of the mutation was facilitated through our technology's superior characteristics:

- full exon coverage
- copy number variants analysis
- high accuracy and resolution

Adventia was able to provide a diagnosis to the family, which enables them to proceed with future family planning knowing all their reproductive choices, such as prenatal diagnosis and PGT-M, to reduce the risk of having another affected child.

# **TECHNOLOGICAL** ADVANTAGES

## TARGETED TECHNOLOGY

Adventia carrier screening 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 Adventia.

## FULL COVERAGE

Adventia screens for **all coding regions*** on the genes of interest, and targets adjacent non-coding sequence, increasing the chances of identifying any pathogenic or likely pathogenic mutations:

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

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

# BENEFITS OF **Adventia**

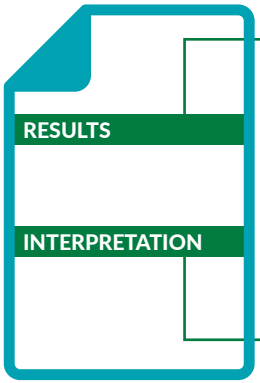
**SAFE**
Non-invasive sample collection

**VALUABLE**
Informs, guides and empowers

**EXPANDED**
Testing up to 231 genes

**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.
The patient is a carrier of the associated disorder.
Sometimes, two disease-causing variants in the same gene may be found. In this case, the patient is either affected now or may be affected in the future by the associated disorder.

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

Summary of the results and recommendations

*Variants of unknown significance (VUS) are not reported as per joint statement of ACMG, ACOG, NSGC, Perinatal Quality Foundation and SMFM[3].*

# WHAT CAN I DO AFTER **Adventia**?

Depending on the results, there are multiple choices you can recommend to your patients:

- Genetic counseling, if applicable
- Prenatal diagnosis during pregnancy
- IVF and preimplantation genetic testing for monogenic disorders (PGT-M)
- Choice of compatible gamete donor without the same mutation
- Early intervention, therapies where available and better clinical management for affected children

**Exceptions apply*

# HOW TO ADMINISTER **Adventia** CARRIER SCREENING?

Recommend **Adventia** 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

The results will be sent to you within 2-3 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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