WHY GET TESTED?

1 in 4 people is a carrier of a genetic disease
Lazarin et al., 2013

Anyone can carry certain mutations (genetic changes) in their body. Some mutations may have no effect on our health and development, while others can cause a genetic disease. When an individual has a mutation in one of their genes, but the mutation is not powerful enough to be expressed, that individual is a **carrier** of a **recessive disease.** Two carriers of the same recessive disease can have a child who is affected, if the child inherits the mutation from **both of them**.

As carriers are **asymptomatic,** they are **unaware of their carrier status** and the risk of passing a mutation to their children. In fact, many mutations for recessive diseases could be inherited via multiple generations without clinical manifestation. Unless you have been tested, it is impossible to know whether you are a carrier of a recessive disease.

Thus, knowing your carrier status can provide information regarding your **reproductive options** and **minimize the risk of transmitting a genetic disease to your children.**

International genetic organizations like the American College of Obstetrics and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend that carrier screening is offered to all people planning to start a family.

DISEASE	CARRIER FREQUENCY	POPULATION
Cystic fibrosis	1 in 45	General population
Alpha thalassemia	1 in 25	General population
Beta thalassemia	1 in 28	Mediterranean
Spinal muscular atrophy	1 in 35	Caucasian

HOW IS **Adventia** ADMINISTERED?

ASK YOUR HEALTHCARE PROVIDER ABOUT Adventia

YOUR HEALTHCARE PROVIDER WILL COLLECT A CHEEK SWAB SAMPLE FROM YOU

THE SAMPLE WILL BE SENT TO OUR LABORATORY

THE SAMPLE WILL BE ANALYZED IN OUR LABORATORY

RESULTS WILL BE SENT TO YOUR HEALTHCARE PROVIDER WITHIN 2-3 WEEKS

MORE **QUESTIONS**?

If you have additional questions or concerns, please ask your doctor. You can also contact us:

NIPD Genetics Public Company Ltd
www.nipd.com
info@nipd.com

NIPD GENETICS

Adventia

carrier screening

Genetic insight to empower your life decisions

SAFE | SENSITIVE | RELIABLE

WHAT IS **Adventia** CARRIER SCREENING?

Adventia is a new genetic test for **carrier screening.** Adventia can be done by any individual to check if they are **carriers** of a genetic disease to minimize their risk of transmitting the disease to their children**.** Carriers are not affected, and don't show any symptoms of a disease, but could have a child who is **affected.**

Adventia PANELS

1 FOCUS PANELS

Six individual panels for **high frequency** and **severe** genetic diseases: *A-Thalassemia, B-Haemoglobinopathies, Cystic Fibrosis, Duchenne Muscular Dystrophy, Fragile X, Spinal Muscular Atrophy.*

19 GUIDELINES BASED PANEL

Single panel that detects 19 genetic diseases **recommended** by international genetic organizations like ACMG and ACOG† due to their **high incidence and severity.** It includes all diseases tested in the Focus panels, and others like *Fanconi Anemia Group C, Phenylketonuria* and *Tay-Sachs disease.*

229 COMPREHENSIVE PANEL

Single panel for 229 diseases that have **moderate to severe,** well-defined phenotype and **high cumulative frequency.** The Comprehensive panel includes all diseases of the Guidelines Based panel, and covers a wide range of metabolic, cardiovascular and haematological diseases amongst others.

For a complete list of the diseases tested by Adventia Guidelines Based and Comprehensive panels please visit **www.nipd.com/adventiapanels**

HOW DOES **Adventia** CARRIER SCREENING HELP ME?

Adventia carrier screening can help you minimize your risk of transmitting a genetic disease to your children, and give you insight into your reproductive choices. By testing moderate to severe diseases that could severely affect quality of life, Adventia can inform you of your choices if a genetic change is identified:

- Genetic counselling on the potential impact of the disease and your reproductive options
- Prenatal diagnosis during pregnancy to know whether your baby is affected
- In-vitro fertilization (IVF) and preimplantation genetic testing (PGT-M) to make sure your baby will not be affected
- Fertility treatments and choosing a compatible gamete donor without the same mutation
- Early intervention, therapies where available and better clinical management for affected children

WHO IS **Adventia** CARRIER SCREENING FOR?

- Couples planning to start their families and want to know about their carrier status
- Any individual or couple going through assisted reproduction, including 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 disease
- High-risk population groups for specific diseases
- People with a family history of a genetic mutation
- Any individual wishing to know more about their genetic background

WHY CHOOSE **Adventia** CARRIER SCREENING?

Adventia was specifically designed to be a **beneficial** and **comprehensive** test for everyone, regardless of ethnic background and family history. It is based on a **novel** and **powerful technology** and can provide **meaningful results** in a short turn-around time to help you minimize your risk of transmitting a genetic disease to your children. The diseases tested by Adventia:

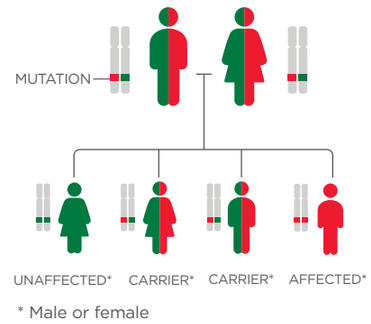
- have moderate to severe phenotype (characteristics)
- are high in carrier frequency
- can severely compromise quality of life
- may be manageable through early interventions

WHAT DOES **Adventia** CARRIER SCREENING TEST FOR?

Adventia screens for **autosomal recessive** and **X-linked** diseases. Carriers of recessive diseases have one healthy gene and one gene with the mutation.

UNAFFECTED CARRIER AFFECTED

Autosomal Recessive Diseases

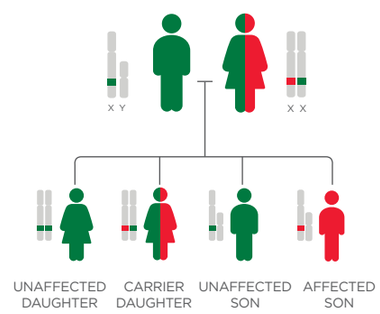


* Male or female

Autosomal Recessive Diseases affect chromosome pairs 1 to 22. If both parents are carriers, they have:

- 1 in 4 chance of having an unaffected child .
- 1 in 2 chance of having a child who is also a carrier, who has inherited the mutation from only one parent.
- 1 in 4 chance of having an affected child, who has inherited mutations from both parents.

X-Linked Diseases



X-Linked Diseases affect the X chromosome, found on the 23rd chromosome pair which determines gender.

- 1 in 2 chance of having a carrier daughter. Female carriers may or may not exhibit disease characteristics due to X-inactivation*.
- 1 in 2 chance of having an affected son. Males who have inherited the mutation are always affected, as they only have one X chromosome.

* X-inactivation is the process of randomly 'silencing' one of the two X chromosomes in females to avoid having double the 'dosage' of protein-coding genes.

WHEN SHOULD I GET TESTED?

Adventia carrier screening can be performed by **any individual or couple** when they wish to learn more about their genetic information to minimize the risk of transmitting a genetic disease to their children. Adventia can also be done **during pregnancy** if prospective parents wish to know whether their child has a risk of having a genetic disease.

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