Genescreen

Empower Your Family's Future: Screen Smart, Plan Safe

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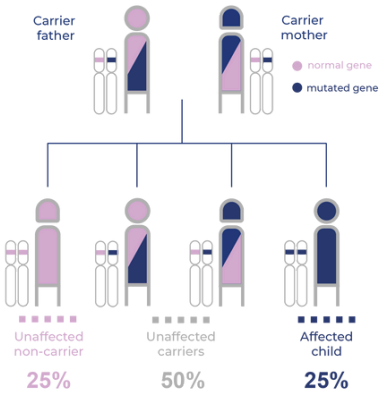
HEALTHY CARRIERS OF GENETIC DISEASES ARE INDIVIDUALS WITH NO OVERT PHENOTYPE BUT CARRYING GENETIC VARIANT(S) WITHIN A DISEASE-GENE.

2-5% of couples are at increasing risk to have an affected baby.[1]

30/10,000 live births are affected by autosomal recessive (AR) and X-linked (XR) diseases.[1]

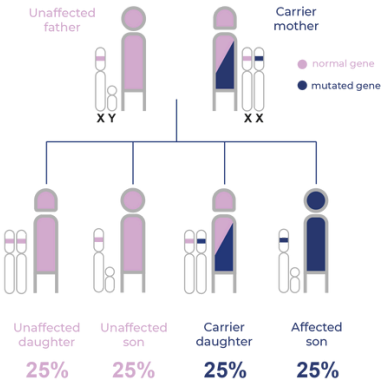
Autosomal Recessive Inheritance:
both parents are carriers



- Both females and males may be affected or carriers **(no sex ratio)**
- **Affected people have two mutations**: one on each copy of the gene (homozygous or compound heterozygous)
- **Carriers have only one mutation** (heterozygous)
- Both parents heterozygous (carriers): **1/4 (25%) risk of affected child**

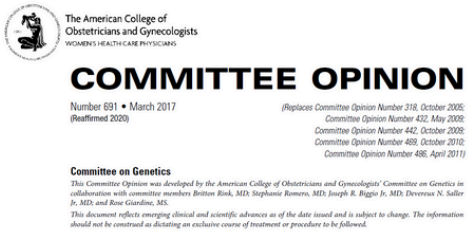
- **Males are affected while females are carriers**
- **Affected males have one mutation on their X chromosome** (hemizygous)
- **Carrier mothers have only one mutation** (heterozygous)
- Carrier mother: **1/4 (25%) risk of affected child (50% of males)**
- **Only females will be tested for XL conditions**

X-Linked Recessive Inheritance:
carrier mother



Carrier screening test enables to identify couples at an elevated risk of having affected pregnancies.

Carrier screening (CS) is a genetic test applicable to individuals and couples within their reproductive age. This screening involves examining disease-causing pathogenic variants in autosomal genes for both members of the couple (for autosomal recessive diseases) or in a gene on the X chromosome for the female partner (for X-linked recessive diseases).


The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

COMMITTEE OPINION

Number 691 • March 2017
(Reaffirmed 2020)

(Replaces Committee Opinion Number 318, October 2005; Committee Opinion Number 432, May 2009; Committee Opinion Number 442, October 2009; Committee Opinion Number 469, October 2010; Committee Opinion Number 486, April 2011)

Committee on Genetics

This Committee Opinion was developed by the American College of Obstetricians and Gynecologists' Committee on Genetics in collaboration with committee members Britton Rink, MD, Stephanie Romero, MD, Joseph R. Biggio Jr, MD, Devereux N. Saller Jr, MD, and Rose Giardine, MS.

This document reflects emerging clinical and scientific advances as of the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed.

"Carrier screening and genetic counseling ideally should be performed before pregnancy because this enables couples to learn about their reproductive risk and consider the most complete range of reproductive options."
ACOG Clinical Guidelines

Genescreen is the carrier screening test developed by Eurofins Genoma and conducted **using Next Generation Sequencing.**

Selection of conditions and genes is based on:

- ***CARRIER FREQUENCY***
- ***PENETRANCE***
- ***CLINICAL CORRELATION OF PHENOTYPE TO GENOTYPE***
- ***GUIDELINES OF MAJOR HUMAN GENETICS AND REPRODUCTIVE SOCIETIES***

- **Cystic Fibrosis (CFTR)**
- **Spinal muscular atrophy (SMN1)**
- **Fragile-X syndrome (FMR1)**
- **α-Thalassemia (HBA1)**
- **α-Thalassemia (HBA2)**
- **β-thalassemia and Sickle cell anemia (HBB)**
- **Xeroderma pigmentosum (XPC)**
- **Oculocutaneous albinism type 1A and type 1B (TYR)**
- **Phenylketonuria (PAH)**
- **Nonsyndromic hearing loss recessive 1A (GJB2)**

PANELS AVAILABLE	Focus	Protect	Easy-Donor	Complete
N genes analysed	>30	>120	>400	>2000
N pathologies analysed	>30 AR/XL	>140 AR/XL	>450 AR/XL	>2100 AR/XL
Who is the test indicated for?	General couples	General couples	Donors	Consanguineous pregnancies couples with family or medical history
		Panel designed on: ACMG Practice Resource[2] SEF Recommandation[3] ACOG Guidelines[4]	Panel alligned with the IVF program of different Countries	Compatible with the majority of panels currently available in the market

CARRIER TEST CUSTOMIZABLE ACCORDING TO SPECIFIC NEEDS

Matching

The option to compare screening test results between the two partners (or donor and recipient), assessing the risk of inheriting diseases in the future child.

Extension

In case the carrier test performed by the partner/donor is not the same as one's own test, it is possible to request an extension of the test to a more comprehensive panel.

Customization

Flexibility to personalize the panel based on patient/couple needs.

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When is the test indicated?

- During or before pregnancy (pre-conceptional or prenatal testing).
- During the reproductive age: all women and their reproductive partners, as well as to gamete (egg or sperm) donors.
  Partners may be tested at the same time or at different moments after one of them tested positive (contextual or sequential approach).
- Consanguinity and positive family history are indications for pre-conceptional/prenatal carrier screening but genetic counselling is recommended.

How to perform Genescreen tests?

Sample collection of BLOOD

Shipping to Eurofins Genoma Lab

DNA Analysis through NGS

Detailed Test Report IN 20 WORKING DAYS

Consult the complete list of analysed genes on the website www.familyproject.it

References:

1. Capalbo A, at al. Considerations on the use of carrier screening testing in human reproduction: comparison between recommendations from the Italian Society of Human Genetics and other international societies. J Assist Reprod Genet. 2022 Nov;39(11):2581-2593. doi: 10.1007/s10815-022-02653-3. Epub 2022 Nov 12.
2. Miller DT, Lee K, Abul-Husn NS, Amendola LM, Brothers K, Chung WK, Gollob MH, Gordon AS, Harrison SM, Hershberger RE, Klein TE, Richards CS, Stewart DR, Martin CL; ACMG Secondary Findings Working Group. ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023 Aug;25(8):100866. doi: 10.1016/j.gim.2023.100866. Epub 2023 Jun 22.
3. Abellàn-Garcìa Sanchèz F., Alamà Faubel P., Aura Masip M., Bassas Arnau L., Castilla Alcalà J.A., Clùa Obrado E., De La Fluente Hernàndez L.A., Guillèn Quilez J.J., Manau Trullàs D., Ruenda J., Ruiz Jorro M., Vendrell Montòn X., Cribado genético en DONACIÓN de GAMETOS. SEF
4. Genetic carrier screening. Royal Australian and New Zealand College of Obstetricians and Gynaecologists, RANZCOG. 2022.

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