Powered With

Next Generation Sequencing (NGS) Technology

GENETIKS 180

Number of genes analyzed

427

Number of diseases assessed

>500

GENETIKS 360

Number of genes analyzed

~22000

Number of diseases assessed

>7000

Empowering Future Health:
Advanced Carrier Screening
for Generational Well-being.

Building Healthy Futures: Safeguarding Generations from Genetic Disorders

Prevention of genetic disorders has a key importance for the generation of healthy families. GENETIKS 180 and GENETIKS 360 provides the broadest carrier screening tests including both common genetic disorders such as SMA, Cystic Fibrosis, Fragile-X, DMD, Thalassemia and rare genetic disorders.

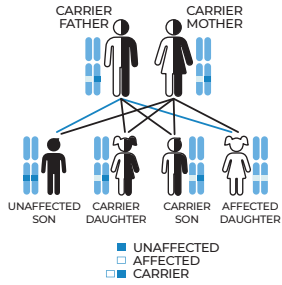
What is a genetic screening test?

It is a test that determines whether or not an individual carries a change in one of their genes and it provides information if they are at increased risk of having a child affected with a genetic disorder.

What is a recessive genetic disease?

The term "recessive" indicates that two copies of the mutated gene, one from each parent, are required for the disorder to manifest. Many carriers only discover their status when they marry someone carrying the same disorder, posing a risk of having an affected child. In families with carrier parents, 25% of their children may inherit both alleles, leading to the disorder. The chances of offspring being carriers or healthy non-carriers are 50% and 25%, respectively (see Figure 1).

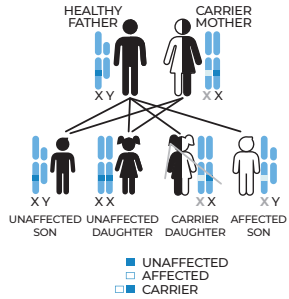
Figure 1: Recessive Inheritance



What is an X-linked genetic disorder?

X-linked inheritance means the gene for a trait or disorder is on the X chromosome. Females have two X chromosomes, males have one. If the mother is a carrier of an X-linked disease, there's a 50% chance her male offspring will be affected. In some cases, female carriers may also show mild or severe forms of the disorder (see Figure 2).

Figure 2: X-linked recessive inheritance



It is estimated that; 400 million individuals worldwide is affected by common or rare genetic disorders.

According to large population studies, one in four individuals (25%) carries a severe genetic disorder. Among isolated ethnic groups and in communities with high consanguinity, couples may share the same genetic mutation, contributing to an elevated likelihood of carrying the same genetic mutation. Leading scientific authorities, including ACOG[a], ESHRE[b], and ESHG[c] recommend carrier screening tests before marriage and childbirth.

[a] *American College of Obstetricians and Gynecologists,*
[b] *European Society of Human Reproduction and Embryology,*
[c] *European Society of Human Genetics*

- **Preconceptional period**
- **Before IVF treatment**
- **Prenatal period**
- **Anyone who wants to identify their risks**

Within the framework of the ACMG guidelines, carrier screening is conducted for over **500** autosomal recessive and X-linked diseases, both common and rare diseases with significant impacts on morbidity and mortality. The panel, guided by the ACMG, covers more than 500 genetic diseases, including SMA[1], Fragile X[2], CFTR[3], DMD[1], and HBB.

- **Preconceptional period**
- **Before IVF treatment**
- **Prenatal period**
- **Consanguineous families**
- **Familial history of a genetic disease**
- **Anyone who wants to identify their risks**

Within the framework of the ACMG guidelines, carrier screening is conducted for over **22,000** genes associated with both common and rare diseases with significant impacts on morbidity and mortality. The panel, guided by the ACMG, covers more than 7,000 genetic diseases, including SMA[1], Fragile X[2], CFTR[3], DMD[1], and HBB.

Variant positivity is confirmed using [1] MLPA, [2] rpPCR, and [3] Sanger sequencing methods.
Please refer to Table 1 for the scope of the panels.

Ten reasons to consider GENETIKS expanded carrier screening tests (180/360)

- **Risk Assessment for Hereditary Diseases:** These tests enable individuals to evaluate their susceptibility to hereditary diseases.
- **Preimplantation Genetic Diagnosis (PGT):** In the preconception phase, the method enables genetic selection in embryos.
- **Prenatal Diagnosis:** The tests allow for prenatal diagnosis during the pregnancy period.
- **Early Diagnosis in Newborns and Children:** Early identification permits the mitigation of harmful effects associated with certain genetic diseases, allowing for intervention through dietary, lifestyle, or necessary treatments.
- **Promoting a Healthy Family:** By offering insights into genetic diseases, the tests contribute to the establishment of a healthier family.
- **Comprehensive Carrier Tests:** Respected authorities recommend screening tests. Aligned with the ACMG Guideline, these tests are among the most comprehensive carrier screenings available. (Please refer to the QR code for detailed panel content.)
- **High Resolution and Coverage Rates:** Leveraging new generation sequencing (NGS) technologies, the tests boast high resolution and coverage rates (>20X 99.6%, 40X 96%, 100X 75%).
- **Gold Standard for Specific Diseases:** In addition to NGS, MLPA, Sanger, and rpPCR methods are employed which are widely accepted as the gold standard for the diagnosis of diseases such as Spinal Muscular Atrophy (SMA), Fragile X, Duchenne Muscular Dystrophy (DMD), Thalassemia, and Cystic Fibrosis.
- **Timely Results:** Results are typically delivered within a maximum timeframe of 4-6 weeks.
- **Flexible Sample Collection:** The tests can be conducted using blood, saliva, or buccal swab (oral epithelial cell) samples.

Table 1: Some of the Single-Gene Diseases Covered in the GENETİKS 180 and 360 Carrier Screening Panels

Alpha Thalassemia (HBA)	Ataxia-Telangiectasia
Autosomal Recessive Polycystic Kidney Disease (PKHD1)	Beta Thalassemia (HBB)
Biotinidase Deficiency	Charcot-Marie-Tooth Disease (PMP22)
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (CYP21A2)	Cystic Fibrosis (CFTR)
Duchenne Muscular Dystrophy (DMD)	Ehlers-Danlos-Like Syndrome Due to Tenascin-X Deficiency
Familial Hyperinsulinism	Familial Mediterranean Fever (FMF)
Fragile X Syndrome (FMR1)	Fraser Syndrome
Gaucher Disease	Gitelman Syndrome
Glutaric Aciduria, Type I	Hemophilia A (F8)
Hereditary Fructose Intolerance	Homocystinuria due to Cystathionine Beta-Synthase Deficiency
Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	Metachromatic Leukodystrophy
Methylmalonic Aciduria - Methylmalonyl-CoA Mutase Deficiency	Non-Syndromic Hearing Loss Associated with GJB2
Phenylalanine Hydroxylase Deficiency (Phenylketonuria - PAH)	Pompe Disease
Retinitis Pigmentosa	Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
Sickle Cell Disease (HBB)	SMA (Spinal Muscular Atrophy)
Spinocerebellar Ataxia 10	Tyrosinemia, Type 1

METHODOLOGY/VALIDATION

Sequence variants and small insertions/deletions:
Whole gene sequences (coding regions and adjacent intronic/splice regions) are covered by >99% of the bases with at least 20 independent sequence reads (20x) by using advanced NGS methods. Intronic and promoter mutations identified in ClinVar and Human Gene Mutation Database (HGMD) are targeted with >98% sensitivity.

Deletions/duplications (del/dup):
Copy number variants (deletions/duplications) are detected using advanced bioinformatics algorithms.

Pathogenic variants:
Pathogenic variants are confirmed additionally by Sanger sequencing, MLPA or quantitative PCR (qPCR) methods depending on the complexity of the genomic region.

Spinal Muscular Atrophy:
Copy number changes (CNV) in the SMN1 gene are screened by NGS and confirmed by multiple annealing looped primed amplification (MLPA) method. Point mutations for spinal muscular atrophy may not be covered due to high sequence homology.

Pseudogenes:
Proprietary bioinformatics tools are used to identify carrier mutations with highly similar inactive counterparts in disease genes (such as GBA for Gaucher disease and HBA1/HBA2 for alpha thalassemia).

REFERENCES:

[1] Lazarin GA, Haque IS, Nazareth S, et al. An empirical estimate of carrier frequencies for 400+ causal Mendelian variants: results from an ethnically diverse clinical sample of 23,453 individuals. Genet Med. 2013 Mar;15(3):178-86. doi: 10.1038/gim.2012.114.

[2]Nguengang Wakap S., Lambert D. M., Olry A., Rodwell C., Gueydan C., Lanneau V., et al. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the orphanet database. *Eur. J. Hum. Genet.* 28 165–173.10.1038/s41431-019-0508-0.

[3]Committee Opinion No. 690: *Carrier Screening in the Age of Genomic Medicine*. Obstet Gynecol. 2017 Mar;129(3):e35-e40. doi: 10.1097/AOG.0000000000001951.

PLEASE SCAN THE QR CODES FOR FULL TEST LIST

ACCREDITATIONS

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Genetiks Genetic Diseases Diagnostic Center is an authorized and licensed laboratory by the Ministry of Health of Turkey.
Genetiks recommends that all genetic tests, requests and follow-ups be performed in a genetic diagnosis center and under the control of medical geneticists.