When Should I Consult a Genetics Center?

1- Do you have any familial relationship with your spouse?
2- Are you and your spouse from the "same" or "nearby" village?
3- In both families:

- Is there anyone with mental disabilities or impairments?
- Is there a disease present in more than one person?
- Is there anyone in your close family who has had multiple miscarriages or children who were born but passed away?
- Is there anyone in the family who is childless?
- Is there anyone experiencing problems during pregnancy?
- Is there anyone with a child who has developmental delay or anomalies?
- Is there anyone diagnosed with cancer before the age of 50? Are there multiple people diagnosed with cancer?
- Is there anyone with hearing loss?
- Is there anyone with severe vision defects, especially at a young age?

4- Do you have an ailment that leads you to visit the doctor multiple times and doesn't seem to improve? (Do you have a chronic illness?)
5- Do you have a disease that cannot be diagnosed or resolved despite treatment?
6- Do you frequently get infections? (pneumonia once a year, more than 2 sinusitis or ear infections per year)
7- Do you have chronic diarrhea?
8- Do you frequently experience cold sores or mouth ulcers, thrush?
9- Have you experienced a decrease in your blood cells?
10- Is there an enlargement of your liver, spleen, or lymph nodes?
11- Have you had severe eczema on your skin?
12- Did your screening for Thalassemia (Mediterranean Anemia) and SMA show carrier status?
13- Have you had an epileptic seizure?
14- Are there any medications you use continuously or a special diet you must follow?
15- Have you had conditions like excessive hair growth or early puberty?
16- Is there a disease said to be genetic in you or your family?
17- If you have had other marriages, did you experience any health problems in pregnancies or children born from those marriages?

If you answered yes to any of these questions, be sure to consult a genetic center.

What Is Pregnarisk and When Should It Be Done?

Are Genetic Diseases a Matter of Fate?

There are about 20,000 genes in the human genome that code for proteins. We all carry certain genetic changes in these genes that can make us carriers for some conditions. For autosomal recessive diseases, both partners must be carriers; for X-linked diseases, the woman must be a carrier; and for autosomal dominant diseases, one partner carrying a disease-related change can create risks for their children.

These risks increase significantly in couples who are relatives, as they often share similar genes and carrier statuses. Even marriages between people from the same or nearby villages can increase the chance of having a child with a genetic condition due to genetic similarity. Large gene panels make it possible to detect many of the genetic changes related to these risks. Couples who wish to identify all possible risks before pregnancy are advised to receive information about these panels.

Identifying risks before pregnancy allows families to take steps toward prevention, making this period especially important. Even when partners are not related, common carrier statuses in the population can still create risks. In dominant conditions, a genetic change carried by a parent may not affect them but may cause health problems in their children. For X-linked diseases, if the mother is a healthy carrier, boys may develop the condition with a 50% likelihood. For these reasons, every couple planning a pregnancy should be informed about these tests.

Although some risks remain due to technical limitations and new genetic changes that can appear for the first time in a child, most inherited risks can be identified, making it possible to prevent many conditions.

Which Tests Can Be Done Before Pregnancy to Ensure a Healthy Baby?

Partners can be tested for carrier status for diseases that are common in the country, severe, or life-threatening. Rare diseases are also important, not only the common ones. Pregnarisk panel tests were created for this purpose.

There is a history of breast cancer in her family and herself.

Marriage Between Relatives (Grandfathers Were Siblings)

There is a family history of heart-related deaths and cardiomyopathy.

Family history includes muscle disease.

There are kidney-related diseases in the family and in the individual.

Muscle disease is present in the family history.

Even if there is no GENETIC disease in the family, we can all carry certain conditions. Couples who are related by blood have a much higher chance of carrying the same gene for a disease.

HOWEVER
Couples who are not related may also carry changes in one or more genes, which can increase the chance of a genetic condition in their baby.

FOR THIS REASON, THIS TEST IS RECOMMENDED FOR ALL COUPLES WHO WANT TO HAVE A CHILD.

İNTERGEN | Genetics and Rare Diseases Diagnosis Center

Health for **Human,**
Science for **Health!**

Panel 1 – Basic Panel

Content of the Panel and Screening Conditions: This panel is recommended for all couples planning a pregnancy or seeking counseling before marriage. The conditions examined in this panel are those that are common in the population, cause very severe clinical symptoms, or lead to early-age deaths. Carrier screening before pregnancy and planning IVF procedures with genetic testing when necessary can help reduce risks in future pregnancies.

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Panel 1 includes:

Chromosome Analysis, Alpha and Beta Thalassemia (Mediterranean Anemia) and Sickle Cell Anemia, Fragile X, Spinal Muscular Atrophy (SMA), Phenylketonuria, Biotinidase Deficiency, Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency, Cystic Fibrosis, Duchenne Muscular Dystrophy (DMD)

Panel 3 – Comprehensive Panel

Content of the Panel and Screening Conditions: Couples planning a pregnancy or seeking counseling before marriage are informed about this test when they request a more detailed evaluation for common conditions. This test can examine most genes known to be associated with diseases in humans and offers very high diagnostic power. Because couples may also be at risk for rarer conditions—especially when there is close or distant consanguinity—“Pregnarisk Panel 3” is recommended.

In this panel, karyotype analysis is performed to determine chromosome-related risks, and whole exome sequencing (WES) is carried out for both partners to analyze the most important regions of nearly all protein-coding genes. Additional tests are included for conditions that are difficult to evaluate through these methods. This approach aims to provide the broadest possible screening. Identifying risks beforehand can guide families toward genetic IVF options to reduce the chance of having an affected child, or help plan diagnostic tests during pregnancy or after birth for couples who still want to conceive naturally.

Panel 3 Includes:

Whole Exome Sequencing (WES) for thousands of conditions, Chromosome Analysis, Alpha Thalassemia, Fragile X, Spinal Muscular Atrophy (SMA), Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency, Duchenne Muscular Dystrophy (DMD), Friedreich Ataxia.

Panel 6 – Extensive Panel

Content of the Panel and Screening Conditions: Panel 6 is a broad screening approach similar to Panel 3 but uses whole genome sequencing (WGS) instead of whole exome sequencing (WES). WGS is a technique that examines almost the entire genome, helping identify genetic variants and supporting both clinical diagnosis and research. While WES focuses on protein-coding regions where most known variants are located, WGS also evaluates additional genetic changes, increasing the potential for identifying disease-causing variants.

Panel 6 includes:

Whole genome sequencing (WGS) for thousands of diseases, Chromosome Analysis, Alpha Thalassemia, Fragile X, Spinal Muscular Atrophy (SMA), Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency, Duchenne Muscular Dystrophy (DMD), Friedreich’s Ataxia.

Genetic disorders are severe, long-lasting conditions that significantly influence the lives of patients and their families. While early diagnosis after birth is essential, preventing these conditions beforehand is the most effective step. Since genetic disorders vary widely, relying on one method alone is not enough to reduce risks. Testing should be tailored to the family’s unique risk factors.

Scan the QR code for the panel contents.

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