INTERGEN | Genetics and Rare Diseases Diagnosis Center

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

How Can You Evaluate Yourself?

In this section, we have created several forms to help you easily answer the question, “When should I consult genetics?” These forms include assessments for autoimmune findings, indicators that may suggest a genetic disease, preliminary evaluation questions for certain cancers, and a form where you can summarize the findings of a child suspected of having a genetic condition. Please complete them as carefully and accurately as possible. If you indicate the presence of any findings in the forms below, you can contact a genetic specialist to receive guidance on the appropriate course of action.

Evaluation Forms

FORM 1

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When Should I Consult Genetics?

1- Do you have consanguinity with your spouse? ◻ Yes ◻ No

2- Are you and your spouse from the same or nearby village? ◻ Yes ◻ No

3- In both families:

- Is there any individual with intellectual disability or disability? ◻ Yes ◻ No
- Is there a disease present in more than one person? ◻ Yes ◻ No
- Are there individuals in your close family with multiple miscarriages or whose child died after birth? ◻ Yes ◻ No
- Is there any individual in the family who does not have children? ◻ Yes ◻ No
- Is there anyone who experienced problems during pregnancy? ◻ Yes ◻ No
- Is there anyone with developmental delay or a child with anomalies? ◻ Yes ◻ No
- Are there individuals diagnosed with cancer before the age of 50? Are there multiple individuals diagnosed with cancer? ◻ Yes ◻ No
- Is there any individual with hearing loss? ◻ Yes ◻ No
- Is there anyone with severe visual impairment, especially at a young age? ◻ Yes ◻ No

4- Do you have a condition that does not improve and causes you to visit doctors multiple times? (Do you have a chronic disease?) ◻ Yes ◻ No

5- Do you have a disease that cannot be diagnosed or does not resolve despite treatment? ◻ Yes ◻ No

6- Do you frequently experience infections? (e.g., pneumonia once a year, sinusitis or ear infections more than twice a year) ◻ Yes ◻ No

7- Do you have chronic diarrhea? ◻ Yes ◻ No

8- Do you frequently have cold sores, oral ulcers, or thrush? ◻ Yes ◻ No

9- Have you ever had low blood cell counts? ◻ Yes ◻ No

10- Is there enlargement in the liver, spleen, or lymph nodes? ◻ Yes ◻ No

11- Have you had severe eczema? ◻ Yes ◻ No

12- Have you been identified as a carrier in screening tests for Thalassemia (Mediterranean Anemia) or SMA? ◻ Yes ◻ No

13- Have you experienced epileptic seizures? ◻ Yes ◻ No

14- Do you regularly use any medication or follow a special diet? ◻ Yes ◻ No

15- Have you experienced excessive hair growth or early puberty? ◻ Yes ◻ No

16- Is there a disease said to be genetic in you or your family? ◻ Yes ◻ No

17- If you had previous marriages, did any health problems occur in those pregnancies or in the children born? ◻ Yes ◻ No

If you answered "Yes" to even one of these questions, you should consult a genetic center.

FORM 2

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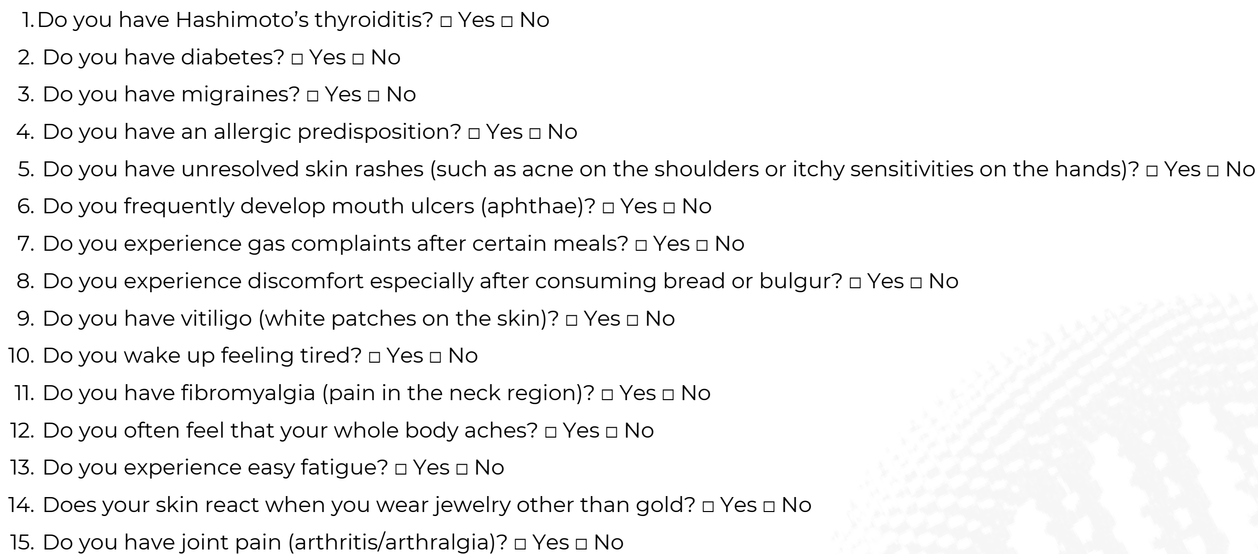
Autoimmune Factors Evaluation Questionnaire



1. Do you have Hashimoto’s thyroiditis? ◻ Yes ◻ No
2. Do you have diabetes? ◻ Yes ◻ No
3. Do you have migraines? ◻ Yes ◻ No
4. Do you have an allergic predisposition? ◻ Yes ◻ No
5. Do you have unresolved skin rashes (such as acne on the shoulders or itchy sensitivities on the hands)? ◻ Yes ◻ No
6. Do you frequently develop mouth ulcers (aphthae)? ◻ Yes ◻ No
7. Do you experience gas complaints after certain meals? ◻ Yes ◻ No
8. Do you experience discomfort especially after consuming bread or bulgur? ◻ Yes ◻ No
9. Do you have vitiligo (white patches on the skin)? ◻ Yes ◻ No
10. Do you wake up feeling tired? ◻ Yes ◻ No
11. Do you have fibromyalgia (pain in the neck region)? ◻ Yes ◻ No
12. Do you often feel that your whole body aches? ◻ Yes ◻ No
13. Do you experience easy fatigue? ◻ Yes ◻ No
14. Does your skin react when you wear jewelry other than gold? ◻ Yes ◻ No
15. Do you have joint pain (arthritis/arthralgia)? ◻ Yes ◻ No

FORM 3

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Liver Cancer Risk Assessment Questionnaire

1. Do you have any diagnosed genetic disease? ◻ Yes ◻ No
2. Is there any individual in your family with liver disease? (e.g., hepatomegaly, fatty liver, elevated liver enzymes, jaundice, etc.) ◻ Yes ◻ No
3. Has anyone in your family or yourself been diagnosed with liver cancer? ◻ Yes ◻ No
4. Is there a diagnosis of cirrhosis in your family or yourself? ◻ Yes ◻ No
5. Is there hepatitis B or hepatitis C carrier status in your family or yourself? ◻ Yes ◻ No

Hepatitis B and C are infectious diseases but do not become chronic in everyone. Therefore, please answer the following questions:

6. Do you have a diagnosis of autoimmune hepatitis? ◻ Yes ◻ No
7. Have you ever been diagnosed with fatty liver (hepatic steatosis)? ◻ Yes ◻ No
8. Have elevated liver function tests (ALT, AST, GGT, etc.) been detected? ◻ Yes ◻ No
9. Have you ever developed jaundice during a febrile illness? ◻ Yes ◻ No
10. Do you frequently experience febrile illnesses? ◻ Yes ◻ No
11. Have you ever had prolonged jaundice or jaundice of unknown cause? ◻ Yes ◻ No
12. Do you or your family have rheumatic diseases, autoimmune diseases, dermatitis, or severe allergic conditions? ◻ Yes ◻ No
13. Do you have diabetes? ◻ Yes ◻ No
14. Do you have a diagnosis of obesity or metabolic syndrome? ◻ Yes ◻ No
15. Do you consume alcohol?

- ◻ No
- ◻ Occasionally
- ◻ Regularly

16. Do you smoke? ◻ Yes ◻ No
17. Have you ever had a blood transfusion? ◻ Yes ◻ No
18. Have you had long-term medication use? (e.g., chemotherapy, immunosuppressive drugs, etc.) ◻ Yes ◻ No
19. Do you or your family have any of the following diseases?

- Hemochromatosis (iron overload) ◻ Yes ◻ No
- Wilson disease ◻ Yes ◻ No
- Alpha-1 antitrypsin deficiency ◻ Yes ◻ No
- Glycogen storage diseases ◻ Yes ◻ No
- Tyrosinemia ◻ Yes ◻ No

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Colorectal (Bowel) Cancer Risk Assessment Questionnaire

1. Is there anyone in your family diagnosed with colorectal (colon or rectal) cancer? ◻ Yes ◻ No
2. Is there anyone in your family who developed colorectal cancer before the age of 50? ◻ Yes ◻ No
3. Has any of the following cancers been seen in your family?

- Endometrial (uterine) cancer ◻ Yes ◻ No
- Gastric (stomach) cancer ◻ Yes ◻ No
- Ovarian cancer ◻ Yes ◻ No
- Pancreatic cancer ◻ Yes ◻ No

(These cancers are important in terms of Lynch syndrome.)

1. Is there anyone in your family with intestinal adenomas (polyps)? ◻ Yes ◻ No
2. Have you ever been diagnosed with a colon polyp? ◻ Yes ◻ No
3. Have you ever been diagnosed with colorectal cancer? ◻ Yes ◻ No
4. Have you ever undergone colonoscopy? ◻ Yes ◻ No
5. Were polyps detected during colonoscopy? ◻ Yes ◻ No
6. Do you have ulcerative colitis? ◻ Yes ◻ No
7. Do you have Crohn’s disease? ◻ Yes ◻ No
8. Have you ever noticed blood in your stool? ◻ Yes ◻ No
9. Have you experienced unexplained weight loss? ◻ Yes ◻ No
10. Do you have anemia (iron deficiency anemia)? ◻ Yes ◻ No
11. Do you have long-standing constipation or diarrhea? ◻ Yes ◻ No
12. Have there been recent changes in your bowel habits? ◻ Yes ◻ No
13. Do you smoke? ◻ Yes ◻ No
14. Do you consume alcohol?

- ◻ No
- ◻ Occasionally
- ◻ Regularly

15. Is your physical activity low? ◻ Yes ◻ No
16. Is your red meat consumption high? ◻ Yes ◻ No
17. Do you frequently consume processed meat (such as sausage, salami, etc.)? ◻ Yes ◻ No
18. Do you consume a high amount of vegetables and fiber-rich foods? ◻ Yes ◻ No
19. Do you have obesity (overweight)? ◻ Yes ◻ No
20. Do you have a diagnosis of type 2 diabetes? ◻ Yes ◻ No
21. Have you ever had a fecal occult blood test? ◻ Yes ◻ No
22. Have you had a colonoscopy within the last 10 years? ◻ Yes ◻ No

Genetic Disease Evaluation Questionnaire in Children

Does the child have any problems in the following areas?

(Multiple selections allowed)

◻ Physical movement (walking, balance, sitting, joint mobility, muscle strength)

◻ Swallowing or feeding

◻ Vision

◻ Hearing

◻ Speech / language development

◻ Toilet control

◻ Learning / attention / behavior

Is there another individual in the family with a similar condition? ◻ Yes ◻ No

Is there any known genetic disease/diagnosis in the family? ◻ Yes ◻ No

Was there any special condition during pregnancy or birth? (Difficult delivery, preterm birth, multiple pregnancy, low birth weight, hypertension, etc.)

◻ Yes ◻ No

Child with Developmental Delay

In which areas is there a significant developmental delay?

◻ Language/speech ◻ Motor (sitting/walking) ◻ Cognitive/learning ◻ Social/communication ◻ Self-care

Developmental Milestones

Head control: ◻ Age-appropriate ◻ Delay

Sitting without support: ◻ Age-appropriate ◻ Delay

Walking: ◻ Age-appropriate ◻ Delay

First word: ◻ Age-appropriate ◻ Delay

Two-word sentences: ◻ Age-appropriate ◻ Delay

Toilet control: ◻ Age-appropriate ◻ Delay

Daily living skills (dressing/using utensils): ◻ Age-appropriate ◻ Delay

Neurological and Behavioral

Seizures/epilepsy: ◻ Yes ◻ No

Balance/coordination problems: ◻ Present ◻ Absent

Behavioral features: ◻ Attention/concentration difficulty ◻ Hyperactivity

Autism-related features: ◻ Limited eye contact ◻ Limited response to name ◻ Repetitive behaviors ◻ Sensory sensitivity

Sensory, Nutrition, Sleep

Suspicion of hearing/vision problems: ◻ Present ◻ Absent ◻ Not tested / unknown

Feeding/swallowing: ◻ Difficulty ◻ Selectivity ◻ Normal

Sleep: ◻ Good ◻ Frequent awakenings ◻ Snoring/suspected apnea

Do behavioral eating/sleep problems affect family life? ◻ Yes ◻ No

Immune / Autoimmune / Allergy

1. Does your child get sick frequently? ◻ Yes ◻ No
2. Does recovery take a long time after illnesses? ◻ Yes ◻ No
3. Are there recurrent ear, throat, lung, or urinary tract infections? ◻ Yes ◻ No
4. Are infections without fever observed (e.g., frequent cough without fever)? ◻ Yes ◻ No
5. Are eczema, atopic dermatitis, or skin rashes present? ◻ Yes ◻ No
6. Are food allergies or symptoms such as abdominal pain, flushing, or irritability after certain foods observed? ◻ Yes ◻ No
7. Are seasonal symptoms (e.g., worsening in spring) noticed? ◻ Yes ◻ No
8. Is there any allergic individual in the family (asthma, allergic rhinitis, eczema)? ◻ Yes ◻ No
9. Are there autoimmune diseases in the family (Hashimoto's, celiac disease, lupus, rheumatoid arthritis, type 1 diabetes)? ◻ Yes ◻ No
10. Does the child have symptoms such as joint pain, swelling, abdominal pain, mouth ulcers, or skin rashes? ◻ Yes ◻ No
11. Are diarrhea, constipation, or abdominal bloating frequent? ◻ Yes ◻ No
12. Are bowel habits irregular? (constipation, diarrhea, irregularity) ◻ Yes ◻ No
13. Are behavioral changes noticed after eating? (irritability, hyperactivity, sleep problems) ◻ Yes ◻ No
14. Do digestive problems occur after antibiotic use? ◻ Yes ◻ No
15. Have behavioral changes been noticed in the child seasonally, after illness, or after vaccination? ◻ Yes ◻ No
16. Does behavior improve or worsen during fever? ◻ Yes ◻ No
17. Is there a history of regression or sudden loss of skills after infections? ◻ Yes ◻ No

If there is a vision problem:

1. Does the child use any visual aid/device? ◻ Yes ◻ No
2. Is it progressive? ◻ Yes ◻ No
3. Are both eyes affected? ◻ Yes ◻ No
4. Is it congenital? ◻ Yes ◻ No
5. Is there nystagmus, a white spot, cataract, or structural eye abnormality? ◻ Yes ◻ No
6. Are there additional problems besides vision? (skin spots, differences in hair or eye color, facial differences, hearing loss, balance problems, delayed walking, epilepsy, developmental delay, or organ problems such as heart, kidney, or liver) ◻ Yes ◻ No

If there is a hearing problem:

7. Is there a cochlear implant? ◻ Yes ◻ No
8. Is there a hearing aid? ◻ Yes ◻ No
9. Is it progressive? ◻ Yes ◻ No
10. Are both ears affected? ◻ Yes ◻ No
11. Is it congenital? ◻ Yes ◻ No
12. Is there a visible ear anomaly? ◻ Yes ◻ No
13. Are there additional problems besides hearing? (vision loss, pigment differences in the eye, balance disorders, heart/kidney problems, growth delay, dental or skin anomalies, etc.) ◻ Yes ◻ No

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