

In which cases is genetic counseling recommended

When there is a family member, newborn child or adult, with:

- a known genetic disorder (gene disorder or chromosomal)
- a possible genetic disorder
- malformations (e.g. dysmorphic features, dysplasia)
- psychomotor or mental retardation

When a couple experiences reproductive problems:

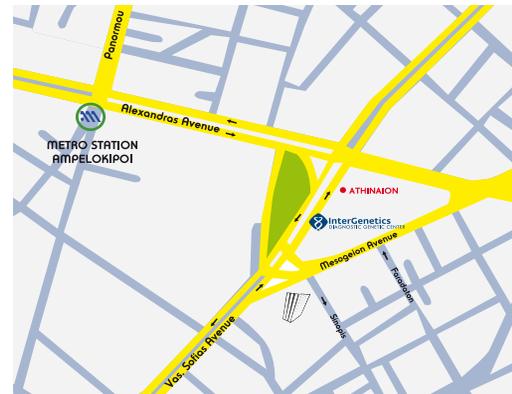
- infertility
- spontaneous abortions
- stillborn children

When during pregnancy, the mother:

- takes medication
- experiences an infection
- is exposed to radiation

To couples, in cases of:

- advanced maternal age
- consanguinity
- an affected family member or with reproductive problems in the wider family (brothers, uncles, etc.)



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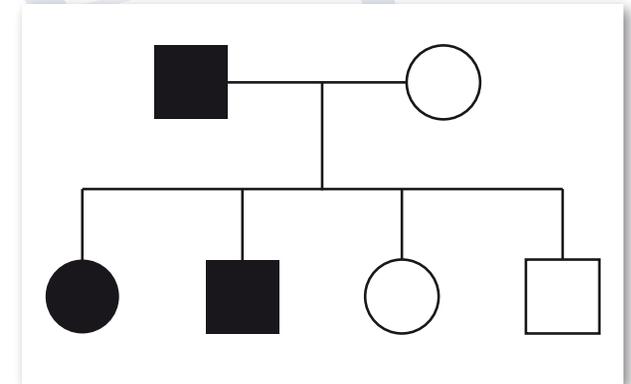
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GENETIC COUNSELING THE FIRST AND LAST STEP OF GENETIC TESTING



- ✓ *substantial experience in clinical genetic evaluation of patients with a pathological phenotype, >9,500 cases*
- ✓ *use of dedicated software tools for in-depth analysis of each case*
- ✓ *close collaboration with the referring clinician and genetic counseling before and after testing*

The prevention of genetic diseases was and still remains the most important way to control them. The identification and application of the laws of heredity is the basis of genetic prevention for families burdened with genetic disorders. Disease prevention in these cases may be reached only after accurate diagnosis of the nature of the disease. Based on current knowledge of how the disease is inherited, the true risks of recurrence within the expanding family and the specific routes of avoiding them are revealed.

This process is called genetic counseling or genetic guidance. Through this, it becomes possible, in many cases, to avoid recurrence of a genetic disease, while it also becomes possible to reduce the anxiety and sense of guilt, which are both unfair burdens for family members.

Main reasons for seeking genetic counseling

- determine the genetic nature of a disease
- disease prognosis for affected individuals and possible treatment
- determining recurrence risks in the family
- possible ways to prevent the disease
- directions for the proper management of the problem



What are the stages of genetic counseling

1. Classification of the disease following:
 - accurate diagnosis through appropriate and specific tests
 - a complete family history
 - investigation of other family members if necessary
 - comparison and evaluation of bibliographic data
2. Calculation of probabilities of recurrence risk in the family, based on known rules of genetics or empirical evaluation
3. Explanation of the data to the individuals
4. Assist the individuals in reaching the appropriate decision
5. Monitoring, consulting and reassessment of the decision, if needed

Reproductive solutions

The reproductive solutions that may be proposed depend not only on scientific-technical issues, but also on other psychological, familial, moral, social, legal, religious and economic factors, which must always be taken into account in each case.

- prenatal diagnosis in the 1st or 2nd trimester
- preimplantation diagnosis via assisted reproduction
- avoiding childbearing (with or without adoption)
- assumption of the risk involved (if the risk is low and the condition has low clinical severity)

Teratogenic agents during pregnancy

- we study the possible harmful effects of drugs, chemicals, radiation and infections for the fetus
- assessment is aided by the use of specific databases at our disposal
- assessment is given in a written report

Note: it is possible to perform evaluation of teratogenic agents in pregnancy remotely. In this case, the referring physician or even the mother may complete the provided specific referral form for teratogens, which can be downloaded from our website, or even sent to you by post or fax. After completion of the assessment by the clinical geneticist, the report will be sent electronically or by fax or by post.