

FIRST LEVEL PRE-TEST GENETIC COUNSELING

Surname: LOMBARDO	CF: LMBBNL60R47H981Y	Proband: 1280.2023
Name: BRUNELLA LOMBARDO		
Date of birth: 07-10-1960	Place of birth: San Marco Argentano	Sex: F
Clinician: COLOMBO LEONARDO	Institute/Hospital: 7	

FAMILIARITY

ZIA

GENETIC COUNSELING

A genetic test is a medical examination that analyzes an individual's DNA to identify changes or mutations that could indicate the presence of or risk for a genetic disorder. In this context, the referring specialist, COLOMBO LEONARDO, has provided the necessary pre-test information and obtained informed consent from the individual, confirming their unequivocal understanding of the documents' contents.

The diagnostic suspect in question is Retinitis Pigmentosa, a group of genetic disorders that affect the retina's ability to respond to light, leading to a progressive loss of vision. Symptoms typically include difficulty seeing at night and a loss of peripheral vision. A genetic test can be particularly useful for this pathology as it may confirm the diagnosis, provide information about the course of the disease, and inform potential treatment options.

From the documentation collected, the criteria for eligibility for genetic testing are met, and the prescription of the same is appropriate. Given that the condition in this case is described as 'FAMILIARE', it suggests a hereditary pattern of Retinitis Pigmentosa within the family. Therefore, there is the possibility of analyzing the relatives of the proband to identify at-risk individuals or carriers and to study the segregation of the genetic mutation within the family.



Sede legale: Ubicazione Esercizio - Conservazione documenti fiscali:
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As per the continuity of care agreement signed with MAGI Laboratory, the specialist will receive the genetic report and will commit to informing the individual about the results of the investigation. If expressly requested in the informed consent, the outcome will also be communicated to the indicated family members. Furthermore, it may be proposed to extend the study to other family members for the identification of subjects at risk or carriers, or for segregation studies.

N.B. All data collected are processed in full compliance with national and European privacy regulations, pursuant to EU Regulation 2016/679 and Legislative Decree 101/2018 and subsequent amendments. Genetic and biometric data will be treated with specific guarantees and protections as required by current legislation, as this information is particularly protected by law.

DATE

CLINICIAN

__06 Dec 2023__
