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| **FIRST LEVEL PRE-TEST GENETIC COUNSELING** | | | |
| Surname: GAITAN | CF: GTNNGB09T69F023T | | Proband: 1185.2023 |
| Name: ANDA GABRIELA GAITAN |
| Date of birth: 29-12-2009 | Place of birth: Massa | | Sex: F |
| Clinician: UNKNOWN | | Institute/Hospital: 7 | |

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| **GENETIC COUNSELING** |
| A genetic test is a medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. In this context, the individual in question has been referred by a specialist whose name remains undisclosed.  Prior to the genetic testing, the individual received two critical documents from the clinician of interest: pre-test information and informed consent. It has been confirmed that the individual unequivocally understood the meaning and implications of these documents, as per the clinician's records.  The diagnostic suspect for this individual is "Distrofia Maculare di Stargardt/Fundus Flavimaculatus," a genetic eye disorder that causes progressive vision loss. This condition typically affects the macula, the central area of the retina responsible for sharp, straight-ahead vision. Symptoms can include blurred or distorted vision, difficulty with fine detail, and gaps or dark spots in the center of vision. The genetic test can be useful for confirming the diagnosis, understanding the progression of the disease, and informing potential treatment strategies.  From the documentation collected, the criteria for eligibility for genetic testing are met, and the prescription of the same is appropriate.  Regarding the genetic nature of the disease, the information on whether the condition is sporadic or familial is not available ('nan'). However, considering the diagnostic suspect, it is important to discuss the possibility of analyzing the relatives of the proband. If the condition is found to be familial, other family members may be at risk, and genetic testing could be proposed to identify carriers or at-risk individuals, or for segregation studies.  As per the continuity of care agreement signed with the MAGI Laboratory, the specialist will receive the genetic report and will commit to informing the individual about the results of the investigation. Unless expressly requested by the MAGI Laboratory, the specialist will also communicate the outcome to the family members specified in the informed consent if explicitly requested. Furthermore, it may be proposed to extend the study to other relatives for the identification of at-risk subjects or carriers, or for segregation studies.  Lastly, it is important to note that all data collected are treated with the utmost respect for national and European privacy regulations, in accordance with EU Regulation 2016/679 and Legislative Decree 101/2018 and subsequent amendments. Genetic and biometric data will be processed with specific guarantees and protections as required by current legislation, as this information is particularly protected by law. |

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