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IN VITRO METHOD FOR THE DIAGNOSIS AND/OR PROGNOSIS OF MALIGNANT MELANOMA

A group of single nucleotide polymorphisms (SNPs) related to the risk and aggressiveness of malignant melanoma (MM) has been identified. Thus, an *in vitro* method based on these biomarkers identification is proposed for diagnosis, prognosis and/or analysis of treatment response of MM.

In the same way, it has been proved with *in vitro* and *in silico* expression assays that these SNPs for risk and aggressiveness of MM are reinforced.

Technology for Licensing

Keywords:

SNP, expression patterns, single nucleotide polymorphism, melanoma, malignant melanoma, biomarkers, CLPTM1L, TYR, precision medicine.

Description:

Malignant melanoma, with a growing prevalence in recent years, is skin cancer's most aggressive form, representing the leading cause of death from skin diseases.

The intratumoral heterogeneity and the complexity of the current status of germline mutations in this type of cancer make it complex to design therapeutic strategies. Thereby, there is a medical need to find reliable biomarkers for an early diagnosis and prognosis of MM, which will facilitate adequate monitoring of the disease, a correct choice of treatment and the control of its subsequent response.

In this sense, the present invention proposes eight risk SNPs related to melanoma and its use as biomarkers and gene signatures of this condition, aiding earlier identification and thus earlier start of the treatment.

These promising markers form the basis of this *in vitro* method that allows a diagnosis of MM in patients at risk or with symptoms from a simple biological sample. As well, it assesses whether patients who are being treated respond to such therapy. Thus, the presence of at least one of these SNPs: CLPTM1L (rs401681), LOC107987026 (rs1335510), TYRP1 (rs2733832), MTAP (rs7023329), TYR (rs1126809), MX2 (rs45430), CTXND2 (rs7412746) and PARP1 (rs3219090); or a high expression level in the TYRP1 and TYR genes point out that the individual has MM, has a poor prognosis or is responding poorly to treatment.

Therefore, this method is an aid to improve precision medicine for MM patients and their closest relatives.

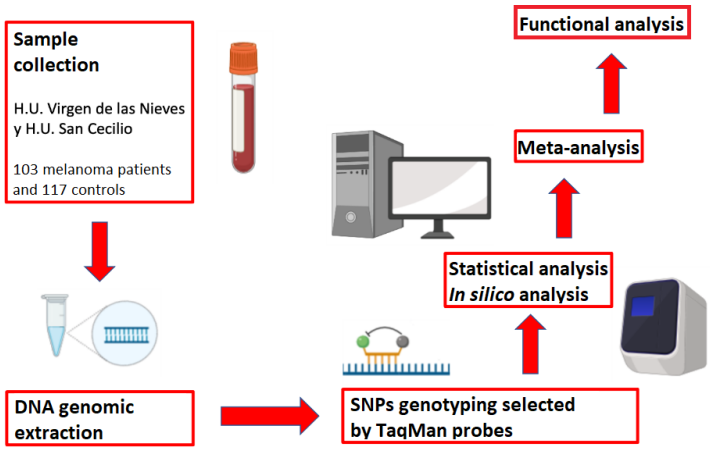
Advantages and Benefits

- Early diagnosis of MM and its prognosis
- Assessment of response to treatment in patients diagnosed with MM
- Improved precision medicine

  It helps to be more accurate in diagnosing melanoma patients and their closest relatives
- The biological sample to be analyzed is easily accessible: skin tissue, saliva, blood, urine, serum or plasma
- No specific equipment is required

  SNPs genotyping and the quantification of the expression levels of these biomarkers is carried out by PCR and/or qPCR
- Simple method

Scheme of the *in vitro* method of this invention



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