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METHOD FOR DIAGNOSING HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)

A new molecular diagnosis method of Hereditary Hemorrhagic Telangiectasia (HHT) has been developed. It is based on the quantification of a set of microRNAs (miRNAs) transported by plasma exosomes. This set also allows diagnosing different variants of the disease.

Since the current methods are not valid for all HHT patients, this method provides new biomarkers for a rapid and reliable HHT diagnosis.

Technology for Licensing

Keywords:

Hereditary Hemorrhagic Telangiectasia, HHT, Rendu-Osler-Weber syndrome, rare disease, molecular diagnostics, exosomes, biomarkers, microRNA, liquid biopsy.

Description:

HHT is a rare disease (estimated prevalence is 1-5/10.000) with an autosomal dominant inheritance that results in arteriovenous dilatations in the skin, mucous membranes and internal organs, with a propensity to hemorrhagic episodes.

The current diagnosis is clinical, but the diagnosis is often delayed due to the gradual onset of symptoms. Thus, having a clinical suspicion, the confirmation test available is a mutational genetic analysis. However, the mutational study in the target genes described so far also fails to diagnose all patients (predominance of new mutations in affected individuals and difficulties in the detection of mutations in heterozygosis and non-coding regions of DNA).

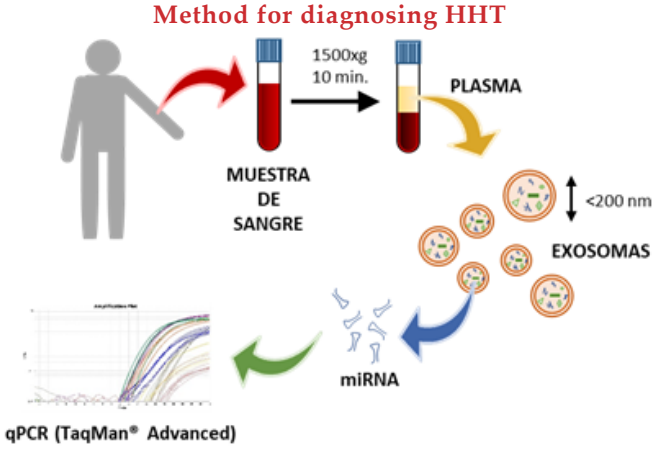
Given this problem, the present invention provides the use of new diagnostic biomarkers, based on the quantification of 8 microRNAs carried by plasma exosomes. These miRNAs provide useful data for the diagnosis of HHT and allow us to classify the disease in two subtypes: HHT1 and HHT2.

Another object of this invention is a kit to assess the expression level of miRNAs through different molecular biology techniques.

Advantages and Benefits

- Early diagnosis
- Independent diagnostic of genetic mutations
- Disease subtypes classification: HHT1, HHT2
- Suitability of the biological sample analyzed:
  - Liquid biopsy (blood extraction): easy to get.
  - Plasma exosomes: high bioavailability and relatively easy to obtain.
- Simple and efficient analysis of disease
- Easy and reliable detection
- Increase of limit of detection and specificity
- The equipment needed is usual and accessible in regular laboratories

Method for diagnosing HHT



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