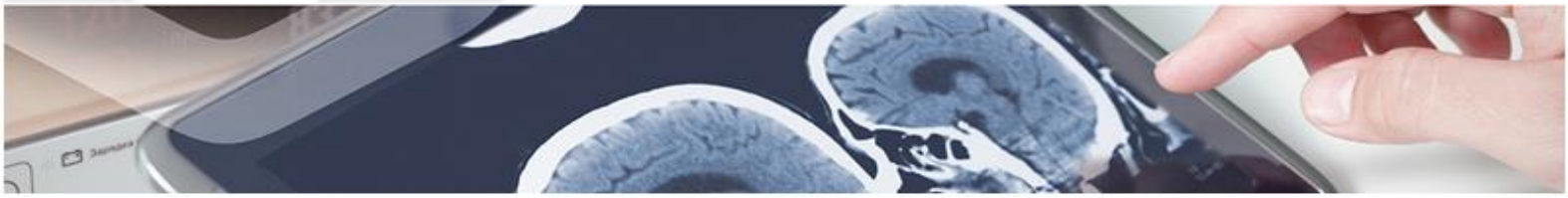
Diagnostics

Molecular Method that radically improves the diagnosis of Hereditary hemorrhagic telangiectasia (HHT).

A research group from Andalusian Public Health System has developed a a diagnostic molecular system for the HHT by getting a liquid biopsy from the patient.

Oficina de TRANSFERENCIA DE TECNOLOGÍA
Sistema Sanitario Público de Andalucía



Description

The HHT or Rendu-Osler-Weber syndrome is a vascular autosomal dominant genetic dysplasia, considered as a rare disease. 85% of the patients present mutations in heterozygosis in the endoglin or in the ALK gene, generating HHT1 and HHT2 respectively, although there are other minority variants.

Currently is used for the HHT diagnosis an agreed clinic profile known as "the Curaçao's diagnostic criteria and they include these four properties. 1) Recurrent epistaxis; 2) Telangiectasias in skin and/or mucosa; 3) Arteriovenous malformation on internal organs; and 4) Family background. The diagnosis of HHT will be made if three of four of these criteria are present. If only one or two criteria are present, it is recommended to determine the genetic mutation, but for technical reasons, it is currently not easy to perform.

This invention allows to analyze the expression patron of the microRNAs that carry the plasmatics exosomes that are HHT characteristic for the fast and easy HHT diagnosis.

Advantanges

Represents a huge advance in the HHT diagnosis respect to the current Curaçao's diagnostic criteria.

It allows to measure the severity of the disease according to the relative expression levels of some of the molecular markers analyzed.

An HHT diagnostic kit or device could be sold for its easy detection anywhere in the world.

Importantly, it allows the diagnosis before the symptoms appear.

Intellectual Property

The technology is protected by a PCT patent application.

Aims

The research group is looking for a collaboration agreement for further development or a licence agreement.

Junta de Andalucía
Consejería de Salud y Familias

Genyo
CENTRO PFIZER-UNIVERSIDAD DE GRANADA-JUNTA DE ANDALUCÍA DE GENÓMICA E INVESTIGACIÓN ONCOLÓGICA

Avda. Américo Vespucio 5, bloque 2, 2ª Plta. Parque Científico y Tecnológico de la Cartuja 41092 Sevilla
☎+34 954 712 376 MÓVIL +34 671 597 655 ☞helena.maldonado@juntadeandalucia.es