Institut d'Investigació Sanitària Illes Balears
IdISBa

Innovation Portfolio

METHOD FOR DETECTING DUPLICATIONS AND/OR DELETIONS IN THE CHROMOSOMIC REGION 22q11.2

INVENTORS: Servei de Salut de les Illes Balears

HIGHLIGHTS

- ✓ **The method has been validated**
- ✓ **The kit developed is cheaper and faster than other genetic techniques**
- ✓ **It can be applied in different pre- and post-natal tests.**

TECH STATUS

- ✓ **TRL6**
- ✓ **PATENT Priority numbers: P201930159**
- ✓ **Priority date: 25 February 2019**

Problem to be solved

The kit that has been developed improves on current techniques for detecting these genetic anomalies, which can take a long time and are also very expensive.

Background and Technology

One of the best known genetic causes of congenital heart disease as well as DiGeorge syndrome are deletions or duplications of the 22q11.2 region. What happens is that this genetic anomaly can lead to other pathologies such as schizophrenia, moderate mental retardation and/or immunodeficiencies. Currently, in order to know all this, different techniques are used which cost a lot of money and need a lot of time.
Therefore, the team of the genomics group has developed a kit that can make a validated diagnosis of whether this genetic anomaly is present in less than 3 hours and at a very competitive price.

Applications

It is the development of a new diagnostic technique for deletions and duplications of chromosome 22q11.2 causing heart disease with all the associated diseases that it entails, which can be applied in hospitals.

Technology status

The kit is developed and a prototype is available for demonstration.

Patent Application number: EP3933051A1
PCT application No: WO2020/174109

Title: Method for detecting duplications and/or deletions in the chromosomic region 22q11.2

Sole authorship of the Servei de Salut de les Illes Balears.

Market Opportunity

Transferred.

Contact Information:
carlos.enrique@ssib.es – *Health Research Institute of the Balearic Islands (IdISBa)*

The patent was licensed to Cybergene AB in May 2022 after a search for companies interested in the technology.

Contact Information:
carlos.enrique@ssib.es – *Health Research Institute of the Balearic Islands (IdISBa)*