VISUVAR

SATT NORD
Technology Transfer Office

Platform for the analysis and visualization of genetic abnormalities identified by CoDE-Seq™ in patients.

Keywords

- Molecular Diagnostic tool
- Precision Medecine
- Next generation sequencing
- Copy Number Variations (CNV)
- Coding point mutations
- Monogenic diseases
- Mendelian diseases
- Augmented whole-exome sequencing (WES)
- Intellectual disibility
- Obesity

Intellectual Property

- Probes are proprietary
- Data management software is proprietary and protected.

Development Status

- Functional prototype

Partnership

We are looking for a partner who can tap the technology

Technology

The Gold standard today in case of suspicion of Monogenic Disease is to use DNA microarray (aCGH) for CNV detection only.

If the answer is negative a Whole Exome Sequencing (WES) should be practiced for more point mutation identification due to poor CNV detection capability,

Quite often, diagnostic is stopped at the step of DNA microarray, whatever the results are, which most part of the time are not very interpretable.

The sequencing of the whole genome also enables this detection but its clinical use is impossible due to the mass of data to be processed and the cost of the analyzes.

CoDE-Seq™ is based on an augmented WES method using a much higher number of probes distributed uniformly throughout the genome.

CoDE-Seq™ is an optimized molecular diagnosis enabling the accurate detection of both copy number variations (CNV) and coding point mutations.

CoDE-Seq™ not only detect all the CNVs identified by chromosomal DNA microarrays but is able to find additional CNVs due to a better resolution.

CoDE Seq™only target 3% of the whole genome reducing time and costs.

CNVs detection through CoDE-seq, aCGH and WES in 40 participants with aCGH data available over 82 patients with Mendelian obesity and/or intellectual disability has shown that CoDE-seq has detected 181 CNVs, WES has only detected 114 CNVs and aCGH 97.

50% of the genetic diagnosis were explained by pathogenic CNVs and 50% caused by pathogenic point mutations, a finding that as not been made so far and should encourage to systematically look for both genetic events.

Benefits

This technology is:

- ❑ Fast (Only one step)
- ❑ Precise
- ❑ Ultra sensitive
- ❑ Easy to interpret
- ❑ Ultra economic (3/10th of the WGS price)

Applications

- Analysis and visualisation of genetic abnormalities in patients suspected of Mendelian obesity and/or intellectual disability.
- Analysis and visualisation of rare Mendelian diseases,

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