NOVEL APPROACH TO DIAGNOSIS HYPOPHOSPHATASIA USING MICROBIOME

A research group from the Andalusian Health Service, the UGR and CIBER has devised a new method to enhance the diagnosis of hypophosphatasia.

The Need

Hypophosphatasia (HPP) is a rare genetic disorder caused by ALPL gene mutations, leading to dysfunctional tissue-nonspecific alkaline phosphatase (TNSALP) and poor bone mineralization. HPP diagnosis is challenging due to symptom overlap with more prevalent bone disorders, leading to frequent misdiagnosis and inadequate treatment. Moreover, it usually requires a combination of genetic, biochemical and radiographic techniques, not ensuring a proper diagnosis.

The Solution

Inventors has defined specific bacterial species in fecal gut microbiota that disappear in HPP patients versus controls, while others uniquely emerge in HPP. These species are proposed as diagnostic biomarkers, particularly for patients with clinical/biochemical signs but no genetic confirmation.

Innovative Aspects

- The technology offers a differential factor to properly overcome diagnostic and therapeutic issues presented by classical approaches.
- Additionally, the depleted species show potential as probiotics to restore HPP microbiota.
- Different combinations of the species can be used to effectively identify HPP in patients.
- The inventors has additionally devised a kit to easily implement the technology in several clinical situations.

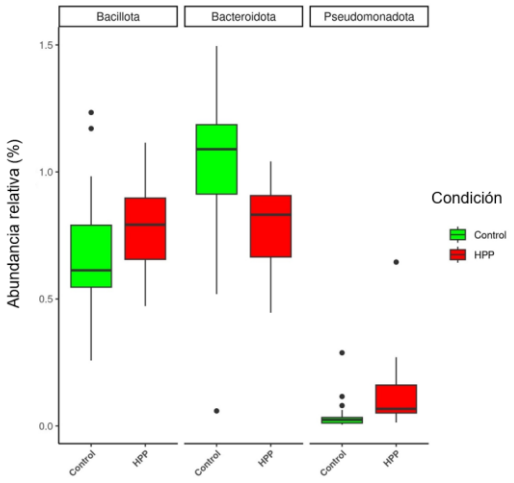

Fig. 1. Example of variations in patient's microbiome

Stage of Development:

The method has been validated with clinical samples as a robust approach to diagnosed HPP with a more accurate procedure and a kit has been designed to easily implement the method in a variety of frameworks.

Intellectual Property:

- Priority patent application filed

Aims

Looking for a partner interested in a license and/or a collaboration agreement to develop and exploit this asset.

Contact details

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