NEW METHOD FOR DIAGNOSING HYPOPHOSPHATASIA

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. To this day, diagnosis requires both persistently low serum alkaline phosphatase (ALP) levels and ALPL mutations, but defining "low" ALP levels is challenging as some patients have near-normal readings. Furthermore, standard sequencing misses non-coding regions and factors mutations, potentially causing underdiagnosis.

The Solution

The research group proposes a novel flow cytometry-based method to detect TNSALP protein levels in blood, offering a fast, cost-effective, and non-invasive diagnostic approach for HPP.

Innovative Aspects

- It is a safe (noninvasive), faster and affordable new diagnostic method.
- It directly measures TNSALP presence, potentially identifying underdiagnosed patients.
- The method provides specific margins and values to identify HPP and to tailor a treatment.
- The method has been implemented in a kit, so it can be easily applied and used in a variety of clinical situations.

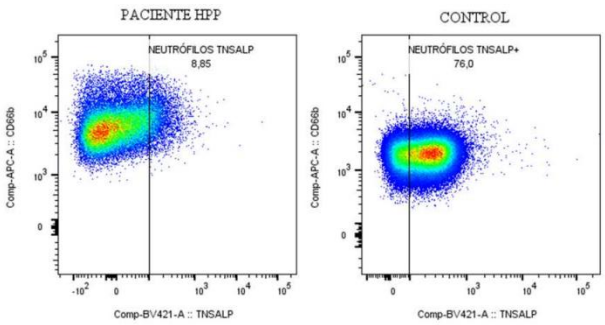

Fig. 1. TNSALP measurement following the proposed method

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.

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