Monitoring of deregulated Acid Ceramidase activity for diagnostics

CSIC and the University of Barcelona have developed a method to measure lysosomal acid ceramidase using a specific substrate and analysis by flow cytometry. This approach avoids interferences with other ceramidases and the use of radioactivity, is highly sensitive (pmol range) and reduces the analysis time to few minutes. Acid ceramidase alteration is directly related to Farber disease and may be relevant in cancer.

Industrial partners from the diagnostics or pharmaceutical industry are being sought to collaborate through a patent licence agreement.

An offer for Patent Licensing

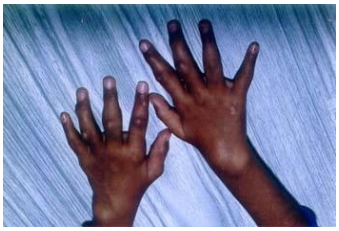
Rapid diagnosis of Farber disease

Acid ceramidase (ACDase) is an enzyme of the metabolism of sphingolipids with a relevant role in maintaining a proper balance in many cell processes.

Farber disease is a rare inherited metabolic disorder caused by ACDase deficiency and storage of undegraded ceramides in lysosomes. Farber disease affects mostly young children, who often die within two years of age. Nowadays, there is no specific therapy but only symptomatic treatment.

Currently, measurement of ACDase can be performed by genetic testing or by biochemical analyses, which require long and laborious analytical methods involving radiolabeling, being only available to highly specialized laboratories.

The method here presented allows easy monitoring of the ACDase activity in lysosomal intact cells by using a specific substrate of ACDase. This compound, once hydrolyzed by the enzyme action and after reaction with a fluorogenic reporter, forms a compound detectable by flow cytometry.



Rapid diagnosis of Farber disease by flow-citometry.

Main innovations and advantages

- Unprecedented flow-cytometry based monitoring of AC in intact cells by means of simple and rapid method.
- High specificity. Since the substrate used is exclusively located at lysosomes, unlike other systems, it is specific for lysosomal acid ceramidase, and no other ceramidases interfere in the analysis.
- Great sensitivity (limit of detection picomoles) and small amount of protein required (10-20 µg).
- Fast time screening (less than 30 min, from sample preparation to results).
- No special equipment and facilities required.
- Useful for diagnosis of Farber disease, cancer or other diseases where the CDase enzyme is deregulated. Moreover, it can be used for screening in investigational drug discovery.

Patent Status

Priority patent application filed

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