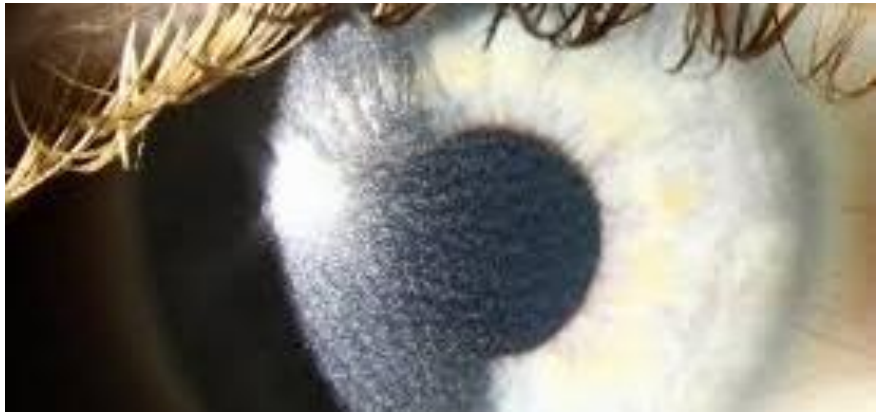
Technology Offer CSIC/IM/061

Rapid diagnosis of diseases related to cystine accumulation



Early and accurate diagnosis and follow up of diseases related to cystine accumulation, such as cystinosis and cystinuria, based on a molecular network producing a signal measurable by fluorescence when certain substances bind to cysteine or cystine, allowing abnormal levels of cystine to be detected.

Intellectual Property

Patent granted in EP and filed in USA (pending)

Stage of development

Validation with cystinosis patients' samples is ongoing

Intended Collaboration

Licensing and/or co-development

Contact

Isabel Masip PhD

Vice-presidency for Innovation and Transfer

isabel.masip@csic.es
comercializacion@csic.es

Market need

Cystinosis is an inherited genetic disease characterized by the accumulation of cystine, leading to irreversible damages in different organs, mainly kidney and cornea. Its only specific treatment is based on the periodic (several times a day) oral administration of cysteamine that reduces the effect of cystine accumulation.

Rapid diagnosis is of utmost importance to initiate treatment as soon as possible to prolong renal function survival and protect extra-renal organs. In addition, the follow up of the intracellular cystine levels for adjustment of the treatment is essential.

CSIC solution

Available methods to quantify cystine levels in leucocytes are based on sophisticated HPLC-MS/MS protocols, which require tedious and elaborated pre-treatment of the sample, only possible in large facilities of reference hospitals with very specialized personnel.

The analytical system presented allows rapid detection of cystine present in blood samples by means of a cheap and reliable fluorescence assay. It may be implemented in any clinical analysis lab, thus allowing preventive screening in prevalence regions and families, and closer monitoring of the patients, improving disease management.

Competitive advantages

- Selectivity. No interference of other biothiols and other typical amino acids commonly present in the sample, overcoming false positives.
- Cystine detection in its reduced (Cys) or oxidized (Cys2) forms without extra preparation steps.
- Applicable to the early diagnosis and follow up of cystinosis and other diseases related to an abnormal cystine storage, such as cystinuria.