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About 15 percent of patients with Wolfram syndrome do not meet current diagnostic criteria

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Researchers at IDIBELL and CIBERER Virginia Nunes and Miguel López de Heredia have collected data from 400 patients with Wolfram syndrome published worldwide in the last fifteen years to better understand the natural history of disease. The findings lead them to propose a revision of the diagnostic criteria of the disease because 15% of paediatric patients escape from diagnosis.

The results of this review have been published this week in the online edition of the journal *Genetics in Medicine* to coincide with the World Day of rare diseases, on February 28th. These rare or minority disorders affect 350 million people worlwide. 80% of these diseases, as Wolfram syndrome, are genetic and half of them are pediatric.

Current criteria

Wolfram syndrome is a recessive a rare, progressive and neurodegenerative disease. It is characterized by the appearance of different diseases in the patient's life: diabetes, optic atrophy, diabetes insipidus, deafness and other nephrological and neurological disorders. Mutations in the WFS1 gene are responsible for these diseases.

Currently, clinical features leading to suspect that a patient suffers Wolfram syndrome are diabetes and optic atrophy in young age (before age 18). The diagnosis is confirmed by genetic analysis.

"Of the 400 patients studied worldwide, 85% showed these two characteristics. This means that 15% of patients are beyond the diagnosis and the treatment will not start until much later", the researcher Virginia Nunes explained. "We believe that if we expand the diagnostic criteria of the disease to the identification of two of the four clinical features of the disease -diabetes mellitus, optic atrophy, diabetes insipidus and deafness-, we would diagnose up to 98% of patients."

Wolfram syndrome Progression

The study made by IDIBELL researchers correlates different WFS1 mutations and clinical characteristics of patients. "We have found different mutations associated with the disease progression. It is the first time we describe the disease progression" explains the leading researcher Miguel López de Heredia.

Lopez de Heredia explains that "this study is only a first approximation. We must keep in mind that we have studied all the publications of Wolfram syndrome patients published during the last 15 years, made with different methodologies and criteria. There is a lack of homogeneity in the data."

Precisely for this reason it is important the work being done in the European and Spanish registres of rare diseases. "There we have all the information," says Nunes, "now we need to work with more data. And one of the problems of the research on rare diseases is to obtain a sufficient number of samples to be useful in the research".

More information: López de Heredia, M. Clèries R. And Nunes, V. Genotypic classification of patients with Wolfram syndrome: insights into the natural history of the disease and correlation with phenotype. Genetics in Medicine 2013, Feb 21. doi:10.1038/gim.2012.18