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New aspects in the evolution of Fabry disease

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Introduction and Aim: The accumulation of globotriaosylceramide (Gb3) caused by a X-linked inherited disorder is called Fabry disease. It is considered one of the most frequent lysosomal storage diseases. More than 1000 mutations of the alpha-galactosidase (GLA) gene are associated with this disease. One of the earliest symptoms in Fabry disease is pain, either episodic crises or chronic pain. Other symptoms may include gastrointestinal, ocular, ear or skeletal manifestations. Cardiac and renal involvements are the most severe complications leading to organ failure and death. The cerebrovascular lesions may be asociated with other severe symptoms that include stroke at younger ages.

The diagnosis of Fabry disease may be put by enzymatic assays of the α-galactosidase A (AGAL-A) activity in plasma or leukocytes but genetic analysis remains the "gold standard" in identifying the precise mutation and even guiding the treatment. Enzyme replacement therapy (ERT) was the first step in treating subjects with this disease and it may decrease the number of sever clinical events and reduce the symptoms. Other therapies include the use of chemical chaperones. This therapy has many advantages including oral administration and was already approved in Europe and US, but it is suitable only for subjects with amenable mutations. Gene therapies (either ex vivo or in vivo) promise to represent a new era for many disorders including Fabry disease, the preliminary data being encouraging.

Although many steps were taken in understanding the pathogeny of Fabry disease, future research is needed especially in the field of therapeutic approaches.

Biography

Robert Dinu has completed his PhD at the age of 28 years from University of Medicine and Pharmacy of Craiova. He has published more than 20 papers in reputed journals and he is the author of many chapters in some books.