Hereditary metabolic diseases

Layachi Chabraoui

Faculté de Médecine et de Pharmacie de Rabat, Ex-Chef du Laboratoire Central de Biochimie et du Centre d'Etude des Maladies Héréditaires du Métabolisme – CHU Ibn Sina Rabat, Maroc.

Abstract

Hereditary metabolic diseases are rare pathologies when considered one by one, but taken together they are relatively frequent diseases. In our experience at the center for the study of hereditary metabolic diseases at the CHU Ibn Sina in Rabat, we were interested in the group of amino-acidopathies and that of lysosomal storage diseases. Among the amino-acidopathies the most common is phenylketonuria (more than 400 cases detected) followed by type 1 thyrosinemia and then homocystinuria. However, other aminoacidopathies such as leucinosis and urea cycle abnormalities are not uncommon. dozens of cases have been diagnosed and the patients folowed up. As far as lysosomal storage diseases are concerned, mucopolysaccharidosis are the most frequently diagnosed with mucopolysaccharidosis type 1 at its head (more than 200 cases diagnosed) followed by Hunter's disease, Morquio disease type A, San -filippo diseases and Maroteaux-Lamy disease. Among sphingolipidoses the most common is Gaucher's disease, but the other pathologies (Niemann-Pick A and B, GM1 and GM2 gangliosidoses, Fabry disease, X-linked adrenoleukodystrophy, metachromatic leukodystrophy, Krabe disease) are not exceptional. .

In conclusion, the collaboration between our team and the pediatricians of the various Moroccan university hospitals has made it possible to develop a notional network of patients care. consequently inherited metabolic diseases are no longer considered orphan diseases.

Biography

Dr Layachi CHABRAOUI
Biologiste médical. Ancien interne des hôpitaux de Lyon. Titulaire des diplômes de spécialités de Biochimie Clinique, Hématologie, Bactériologie et Virologie, Immunologie et Parasitologie.
Ex-Professeur à la Faculté de Médecine et de Pharmacie de Rabat
Ex-Chef du Laboratoire Central de Biochimie et du Centre d'Etude des Maladies Héréditaires du Métabolisme – CHU Ibn Sina Rabat
Président de la Société Marocaine de Chimie Clinique et Biologie Médicale (SMCC)
President of de The Moroccan Society for Study of Inborn Errors of Metabolism (MSSIEM)
Président de la Fédération Internationale Francophone de Biologie Clinique et Médecine de Laboratoire (FIFBCML)
Past-President of the Arab Federation of Clinical Biology (AFCB)"

E-mail: soylak@erciyes.edu.tr