Abstract

This is the first of a series of articles on inborn errors of metabolism, their physiopathology, clinical diagnosis and treatment and the pursuit of treatment options, currently being researched, to upgrade treatment of these patients in the future. In this country, under National State Law No. 24.438 and Santa Fe Provincial State Law No.10.987, tests for the early detection of Phenylketonuria (PKU), inherited hypothyroidism (HC) and cystic fibrosis (FQ) have been enforced. In addition, galactose tests and 17 hydroxyprogesterone by EIA (Enzymeimmunoanalysis) are prescribed for the diagnosis of Galactosemy and Congenital Adrenal Hyperplasia, respectively.