Case Series

Peroxisomal disorders: our experiments and physician guide

Yousef Shafeghati[1] , Fariba Afroozan[2], Navid Almadani[2], Ghazal Vakili[1]

1- Genetics Research Center, University of Welfare Sciences & Rehabilitation, Tehran, Iran

2- Kariminejad - Najmabadi Pathology & Genetics Center, Tehran Iran

Abstract

Peroxisomes are responsible for a number of very important metabolic reactions, including synthesis of glycerol ethers, shortening very long chain fatty acids (VLCFAs; C24:O and C26:O), and oxidation of the side chain of cholesterol needed for bile acid production. Peroxisomal biogenesis disorders (PBDs) are genetically and phenotypically related disorders that involve enzymatic activities of peroxisomes. They are rare mostly autosomal recessive diseases characterized by multi-systemic structural and functional abnormalities. A number of biochemical abnormalities have been described in PBD patients including decreased levels of plasmalogens, and increased levels of VLCFAs and cholestanoic acid derivatives. More than 25 different entities have been diagnosed and reported in the last two decades. The most severe condition is the Zellweger syndrome, a condition due to the absence of functional peroxisomes. Affected patients are severely ill, and show multiple congenital anomalies and neurological aberrations. Chondrodysplasia punctata is another example; they are genetically heterogeneous group of dysplasias having stippling of the epiphyses in infancy as a common feature. Peroxisomal abnormalities only found in the rhizomelic type I. There are specific biochemical tests for evaluating peroxisomal functions. The diagnoses on suspected cases can now be confirmed precisely by detailed biochemical evaluation and molecular analysis in some metabolic centers. Accumulation of certain VLCFAs (C24:0, and C26:0); deficiency of plasmalogens, and elevation of phytanic acid are some of them. Herein we report 10 Iranian families with 15 affected cases of Zellweger syndrome and rhizomelic type I chondrodysplasia punctata (RCDP I).

Key words: Peroxyzomal disorders; Zellweger syndrome; Chondrodysplasia punctata; Very long chain fatty acids; Low-level plasmalogen

***Yousef Shafeghati, MD**
University of Social Welfare Science and Rehabilitation,
Genetics Research Centre, Evin, Tehran, Iran
E-mail: y_shafeghati@yahoo.com/ Tel: +9821 22407814