Leukodystrophies

남 태 승
전남대학교 의과대학 신경과

Tai-Seung Nam, MD, PhD
Department of Neurology, Chonnam National University Medical School

Leukodystrophies are a heterogenous group of inherited progressive neurologic disorders, which primarily affect white matter (WM) by a failure in myelination or hypomyelination. It is characterized by a diffuse demyelination on pathologic examination as well as an extensive central nervous system (CNS) WM signal abnormalities on the magnetic resonance imaging of the brain or spinal cord. Leukodystrophies may be differentiated from acquired medical conditions including the more broader term "leukoencephalopathy" by 1) the existences of genetic causation or familial inheritance, 2) a progressive clinical course, and 3) a predominant WM involvement of CNS.

Leukodystrophies can be classified as early (infantile, or childhood)- and late (juvenile, adult)-onset according to the onset of disease. In general, the clinical presentations of late-onset may be broadly similar to those in early-onset. Leukodystrophy patients may present with CNS dysfunction symptoms and signs including psychomotor slowing, declined cognitive function, spastic limbs, ataxia, bulbar symptoms, or movement disorders, and etc. However, their symptoms may be sometimes considerably variable, atypical, and also overlapped in symptomatology irrespective of the types of leukodystrophies. Further, it can be not easy to assess whether the clinical courses are static or progressive. Therefore, leukodystrophy can be frequently overlooked, or misdiagnosed as having another medical conditions such as inflammatory demyelinating diseases, vascular diseases, toxic, infectious or nutritional disorders, especially in patients with late-onset or acute-onset.

Many types of leukodystrophies have been identified until now. Among them, X-linked adrenoleukodystrophy/Adrenomyeloneuropathy, Alexandeer disease, Adult-onset autosomal dominant leukodystrophy, Canavan disease, Krabbe disease, Metachromatic leukodystrophy, Pelizaeus-Merzbacher disease have been well known in the clinical practice. Recent advances in the radiologic technologies and molecular genetics can help us to verify their etiologies (causative genes), understand pathomechanisms, and classify leukodystrophies.

Today, I want to discuss broadly the clinical and radiologic spectrum of luekodystrophies, particularly in late-onset type.

Key Words: Leukodystrophy; Magnetic resonance imaging; Genes

Tai-Seung Nam, MD, PhD
Chonnam National University Medical School, Gwangju 501-759, Korea
TEL: +82-62-220-6175 FAX: +82-62-236-0839
E-mail: nts0022@hanmail.net