ALS의 진단과 치료: 도전과 과제

이 광 우
서울의대 신경과 명예교수

Diagnosis and Treatment of Amyotrophic Lateral Sclerosis: Achievements and Challenges

Kwang-Woo Lee, MD, PhD
Emeritus Professor, Department of Neurology, Seoul National University College of Medicine

Amyotrophic lateral sclerosis (ALS) is a typical neurodegenerative motor neuron disease characterized by progressive cell death of upper and lower motor neurons. Patients usually die from respiratory failure on average within 3-5 years after symptom onset. More than 150 years have passed since Jean-Martin Charcot first described clinical and pathologic characteristics of the disease, but we still have many unresolved questions about the cause and pathogenesis of the disease and there are still no satisfactory treatments yet. Recently, however, it is becoming increasingly clear that ALS is not a single disease but a syndrome, i.e., a collection of several diseases. Appraisal of this concept will provide an important step towards precise classification of ALS, and is important for the development of effective targeted therapeutics based on the cause and major pathogenic mechanisms of the disease.

In particular, it is well known that there are significant variations in the site of onset (bulbar vs. limb onset), the degree of upper and lower motor neuron dysfunction, cognitive impairment, and the rate of disease progression. In addition, over the past 10 years, more than 25 disease-causing genes have been identified with the development of molecular genetics technology. Recent advancements in biomarkers (biofluid, electrophysiological and neuroimaging) and gene/cell therapy have also provided deeper understanding of the heterogeneity of ALS.

In this lecture, I will provide an overview of the diagnosis and treatment of ALS, focusing on the diversity and heterogeneity of the disease in terms of clinical phenotypes, genotypes and pathological mechanisms. Touching on domestic and international major research results, I will also address how to translate the heterogeneity of ALS into more accurate diagnosis, classification and development of effective targeted therapy.

Kwang-Woo Lee, MD, PhD
Emeritus Professor, Department of Neurology, Seoul National University College of Medicine
E-mail: lkw3215@gachon.ac.kr