National Registry of Designated Intractable Diseases in Japan: Present Status and Future Prospects

Yasuhiro Kanatani\textsuperscript{1,2}, Hiroshi Mizushima\textsuperscript{1,2}

\textsuperscript{1}Director, Department of Health Crisis Management, National Institute of Public Health, \textsuperscript{2}Director, Center for Public Health Informatics, National Institute of Public Health

Recently, more than 6000 rare diseases have been identified (defined as affecting <5/10,000 individuals in Europe and <200,000 people in the United States). In Japan, a certain number of rare diseases were designated intractable diseases for promoting research to clarify the pathogenesis of these diseases and to develop pharmaceutical products and medical devices. The promotion of a clinical study on rare diseases is necessary to construct a national database because demographic as well as genomic data are required for evaluating the prognosis of patients with rare diseases. On the other hand, the diagnostic criteria for rare diseases should be revised according to the progress of medical diagnostic technologies; therefore, the standardization of a registration is necessary for re-thinking the classification and categorization of diseases. The Ministry of Health, Labor, and Welfare (MHLW) in Japan launched an online registry system for gathering designated rare and intractable diseases in 2001. Although the registration rate of these diseases has improved each year, a remarkable difference exists among prefectures. To address these issues, the MHLW established a new Act on intractable diseases that enables registration at public health centers as well as at hospitals that satisfy certain criteria. Herein, we describe the present status of the national registry of designated intractable diseases in Japan and discuss future prospects.