Country: Korea

Organization: Korea National Institute of Health

Program/Project: The national program on rare diseases was launched in 2001 as a subsidy program for medical expenses. The Korea National Institute of Health (hereinafter referred to as 'KNIH') tries to establish clinical networks for rare diseases to collect clinical data for patients, increase knowledge of pathophysiology and natural history of rare diseases, finally diagnose the rare disease. The KNIH developed and established the Korean Mutation Database (KMD, http://kmd.cdc.go.kr), a country-specific database of human gene mutations, in September 2009. Since 2012, has started the genetic testing financial assistance program. Total 1,426 patients were applied for this program and then 1,099 patients were diagnosed. Further analysis is to be done for the rest of the patients. As KNIH is conducting various activities at domestic level, it is also participating in international consortium IRDiRC as a member institution.

So far, a couple of hospitals in Korea have been making their efforts to conduct a diagnosis support program for undiagnosed patients on their own. However, they have experienced some limitations in terms of budget and network. KNIH is planning to conduct a pilot program of UDP in 2017 and in the final process of designating a study center for that program. The initial goal of this program is to start and stabilize Next Generation Sequencing (NGS)-based diagnostic services for undiagnosed rare disease patients. By launching this pilot program with government funding in 2017, we expect that we can set up a continued support systems for patients and their families and those we can contribute to the advancements in medical technology and knowledge.