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Genome project aims to diagnose patients with rare diseases

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In a potential ray of hope for thousands of people with undiagnosed conditions, the Japan Agency for Medical Research and Development has launched a project to refer such patients to a centralized network of specialists for genome analysis.

The Initiative on Rare and Undiagnosed Diseases (IRUD), launched Wednesday, is designed to help people suffering from medically unidentified conditions find a diagnosis and receive expert consultation, taking advantage of advances in genetic testing techniques.

Currently, there are about 7,000 diseases that result from genetic abnormalities that affect nerve and metabolic systems. But because each has only a few patients, doctors are often unable to give an accurate diagnosis, said Kenjiro Kosaki, a professor of medical genetics at Keio University Hospital in Tokyo and the sub-leader of the project.

While there are no official statistics on the number of people in Japan with undiagnosed conditions, Kosaki estimates that more than 10,000 babies are born with such conditions nationwide every year, citing numbers for the U.K., where, with a population roughly half the size of Japan, around 6,000 babies are born annually with unknown syndromes.

“Genome research has had little connection with medical treatment (in Japan) until now,” said Kosaki, who is certified as a medical geneticist in the U.S. “This project is patient-centric, in that we now have a system to deal with patients with a wide range of symptoms.”

The government-funded project will this fiscal year cover up to 1,000 children or adults who had a childhood onset of undiagnosed conditions. Such patients will be referred to one of 17 hospitals around the nation staffed with expert doctors on rare diseases. If a diagnosis is not given at that level, the patients will be referred to one of four designated institutions — the National Center for Child Health and Development (NCCHD) and Keio University, both in Tokyo; Tohoku University in Sendai; and Yokohama City University.

These institutions will carry out genome testing by taking patients’ blood and running them through genome sequencer devices. The sequencing will give trained clinicians clues to identify genetic abnormalities that cause rare diseases, including ones involving

developmental retardation and accompanying physical symptoms in internal organs and limbs, Kosaki said.

The IRUD project, funded by the AMED, a government medical research and development body launched in April, follows the examples of the Undiagnosed Diseases Program by the U.S. National Institutes of Health and the Deciphering Developmental Disorders project in the U.K.

The U.S. UDP, launched in 2008, has received 9,300 inquiries and seen or scheduled 700 patients as of May 2014, according to its website. It has succeeded in resolving between 25 and 50 percent of patients with a clinical, molecular or biochemical diagnosis.

Japan's project will also allow the nation's doctors —who have so far conducted genetic research independently — to deepen their network and information-sharing with geneticists overseas, said Yoichi Matsubara, IRUD project leader and director of NCCHD's research institute.

"International cooperation is active in this field, because some of the extremely rare diseases have only one patient in one country, and there are cases in which a new disease is identified by finding one or two patients in another country," Matsubara said.

The project includes the establishment of a genome database of people with rare diseases.

"Such genome information is extremely important as it can help future sufferers of a rare disease," Matsubara said. "The fewer the patient numbers are for the disease, the more valuable the patients' prognoses will be, such as whether they might have heart disease five or 10 years down the line. We would like to create a solid database of clinical and genetic data of patients involved in the project."

One of the biggest challenges for Japan is the lack of trained physicians who can analyze the sequencer data, Kosaki said, adding that compared with the situation in the U.S., such specialists are still few and far between in Japan.

Keio University Hospital's IRUD team can be contacted at 03-5363-3906, from 9 a.m. to 5 p.m. Monday through Friday or via irud@skip.med.keio.ac.jp (mailto:irud@skip.med.keio.ac.jp) .

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