September 3, 2021
Kobe City Eye Hospital
Sysmex Corporation
RIKEN GENESIS Co., Ltd.

Approval of "Genetic Diagnosis and Counseling for Inherited Retinal Dystrophy" Using Gene Panel Testing System for Advanced Medical Care B

Kobe City Eye Hospital (Kobe, Japan; Director: Yasuo Kurimoto) announces that on September 2, 2021, it obtained approval of "genetic diagnosis and counseling for inherited retinal dystrophy (IRD[1])" using the IRD Panel Testing System (provisional name), a genetic testing system which has been co-developed with Sysmex Corporation (HQ: Kobe, Japan; Chairman and CEO: Hisashi Ietsugu), as Advanced Medical Care B.[2] Kobe City Eye Hospital is planning to provide the testing service from October 2021 or thereafter.

IRD is a collective term for a broad group of hereditary progressive diseases including retinitis pigmentosa, a designated intractable disease that causes partial or complete vision loss. As these diseases progress, patients require daily support.

In Japan, diagnosis and treatment of IRD are currently based on clinical symptoms and ophthalmic examination, however recent research is shedding light on its causative gene. Once the causative gene is identified, the mode of inheritance will be elucidated and prognosis can be predicted, making it possible to not only determine a treatment plan based on the causative gene, but also map out support plans for low vision care[3] and molecular evidence-based genetic counseling that spans the entire life of the patient. If this can be realized, IRD patients' quality of life (QOL) will be enhanced significantly, as they may be able to prepare themselves for careers or make future family plans based on their risk of developing IRD.

So far, pursuant to the Comprehensive Collaboration Agreement signed in February 2020, Kobe City Eye Hospital and Sysmex have been engaging in joint development of the IRD Panel Testing System, a genetic testing system for identifying causative genes in IRD patients ("Testing") to implement genomic medicine for the treatment of patients with hereditary retinal degenerative diseases in clinical settings.

Now that "genetic diagnosis and counseling for IRD" using the Testing have been approved for Advanced Medical Care B as of September 2, 2021, Sysmex will commence the Testing at Kobe City Eye Hospital from October 2021, while at the same time attempting to persuade more institutions engaged in the provision of advanced medical care to provide the Testing in order to expand opportunities for IRD patients to receive it.

The clinical utility of genetic counseling, Testing, planning of treatment and low vision care based on the Testing result and expert consultation (Expert Panel[4]) will be evaluated during as Advanced Medical Care B. Based on the results of evaluation, we aim to apply this IRD clinical flow for coverage

by national health insurance.

The Testing will be provided at the Innovation Genome Center (Kawasaki Office) of RIKEN GENESIS Co., Ltd. (HQ: Shinagawa-ku, Tokyo, Japan; President and CEO: Kenji Iwakabe), a subsidiary of Sysmex capable of assuring the quality and precision of genetic testing.

Kobe City Eye Hospital, Sysmex, and RIKEN GENESIS will continue to work toward the improvement of people's well-being, as well as the advancement of medicine, as we seek to improve patients' QOL by providing this Advanced Medical Care B to achieve early implementation of the clinical flow for IRD.

Reference

Press release dated March 5, 2020: "Sysmex and the Kobe City Eye Hospital Sign a Comprehensive Collaboration Agreement -Reinforcing Collaboration toward the Realization of Genomic Medicine in the Area of Ophthalmic Disorders-
https://www.sysmex.co.jp/en/news/2020/pdf/200305_01_e.pdf

Terminology

1 Inherited Retinal Dystrophy (IRD):
A hereditary progressive disease presumably caused by a gene mutation. Its main symptoms include night blindness (difficulty seeing in dark settings), tunnel vision (a narrowing field of vision), and progressive loss of vision, which can lead to a complete loss of vision in severe cases. Several diseases that present similar symptoms are collectively referred to as IRD. It is estimated that one out of every 4,000 to 8,000 people develops an IRD. The most common IRD subtype is Retinitis Pigmentosa (a designated intractable disease, Notification No: 90).
Disease explanations, Japan Intractable Diseases Information Center (Japanese only):
https://www.nanbyou.or.jp/entry/196

2 Advanced Medical Care B:
Advanced Medical Care refers to novel, promising medical technologies whose efficacy, safety, and other parameters have yet to be well established, and are designated as such by Japan's Ministry of Health, Labour and Welfare to evaluate their potential in order to determine whether they should be covered by national health insurance in the future. As an exception, it is allowed to combine payment for the latest technologies that have not yet been covered by public health insurance and self-pay medical service under the existing public health insurance system (mixed payment). Provision of Advanced Medical Care technologies are limited to hospitals and clinics that meet the institute criteria accepted by the MHLW for each designated technology.

3 Low vision care:
To those who experience any difficulty in their daily lives due to vision problems, a wide variety of support is offered, including the provision of advice or medical devices to improve QOL, provision of various counseling and information services on careers and employment, and use of welfare programs.

4 Expert Panel:
For Advanced Medical Care B that has received approval, a conference comprising ophthalmologists, clinical genetics specialists, certified genetics counselors, bioinformatics specialists, and other experts will be assembled to discuss the Testing result and the treatment plan for the patient.
