Brain Research Institute, Niigata University
SUGIE Atsushi, Associate Professor

Pathogenicity Validation Service for Gene Mutations: Diagnostic Support in Genomic Medicine

【Keywords】

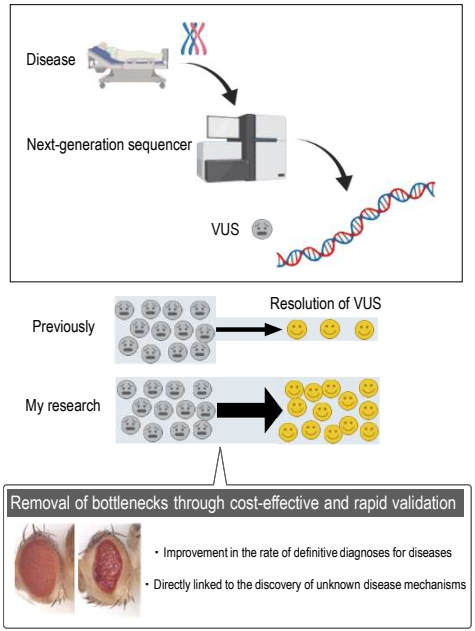
Genetic testing service	Genome medicine	VUS	Hereditary disease	Drosophila

■Summary

Advancements in genetic testing technology have accelerated the analysis of patient DNA, enabling definitive diagnoses for hereditary diseases. However, the growing number of "Variants of Uncertain Significance (VUS)"—genetic mutations with unclear links to diseases—has introduced a new challenge. Traditional methods using mouse models and iPS cells are cost-prohibitive and incapable of resolving the increasing number of VUS. I aim to offer a cost-effective and rapid genetic analysis service using Drosophila, focusing on elucidating these VUS. Initially, this service will target clinical researchers, with plans to expand to hospitals and genetic testing service providers. Furthermore, by collaborating with pharmaceutical companies, it will contribute to the development of new treatments.

■Subject Details/Topic

Our technological seeds are based on research outcomes spun out from the AMED-led initiative, the Initiative on Rare and Undiagnosed Diseases (IRUD), with a core focus on clarifying VUS. IRUD is a large-scale research project that leverages genomic medicine to diagnose patients with rare and undiagnosed diseases. My role involved elucidating the pathogenic significance of VUS identified through genomic analysis. The use of Drosophila in our research provided a simple and rapid method for identifying the pathogenic effects of these variants. Two major advantages of this approach are: 1) not only can we determine whether the variant is disease-related, but we can also identify whether the mutation results in loss of function or gain of function, and 2) it is both cost-effective and fast. Currently, no platform exists that can provide answers for VUS with unknown pathogenic significance. Given the high costs associated with mouse models and iPS cells derived from human patients, our unique technology is expected to occupy a crucial niche in the field of genetic functional analysis in the future.



Sakamoto et al., Hum. Mol. Genet., 2021;
Nitta et al., Hum. Mol. Genet., 2023;
Vetro et al., American Journal of Human Genetics 2023;
Yamada et al., European Journal of Medical Genetics, 2023;
Itai et al., Sci. Rep., 2023;
Iida et al., Frontier in Genetics 2024

■We hope to collaborate with…

Genetic testing services, the pharmaceutical industry, diagnosis and treatment of hereditary diseases, cancer research, etc.

Contact us: Institute for Social Innovation and Cooperation, Niigata University
TEL：025-262-7554 FAX：025-262-7513 E-mail：onestop@adm.niigata-u.ac.jp