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Fujitsu, Kyoto University build AI system to detect genetic disorders

March 15, 2021 | News

The AI verification system called "MGeND Intelligence" estimates the disease-causing potential of genetic mutations



Fujitsu Limited and Kyoto University research group led by Professor Yasushi Okuno of the Kyoto University Graduate School of Medicine, have developed an AI verification system called "MGeND Intelligence".

When the genetic mutation information of a patient is input into this system, its ability to cause disease is estimated by pathogenicity estimation AI using machine learning technology and the explanatory text of the basis of this finding is generated and displayed together with the estimated result by the explainable AI.

This explanation offers a useful reference to doctors who are considering a treatment plan or genomic medical researchers. The system draws from the "Integrated Database of Clinical and Genomic Information program", managed by the Japan Agency for Medical Research and Development, and from April 2021, Kyoto University plans to offer MGeND Intelligence to joint researchers and institutions.

In addition to the pathogenicity estimation AI, the MGeND Intelligence verification system includes explainable AI that shows the basis of the system's findings, as well as literature search AI that supports the retrieval of related articles. Working with the Integrated Database of Clinical and Genomic Information, "MGeND," which was made public by Kyoto University in 2018, Fujitsu supports the research and clinical interpretation of genetic mutations, including mutations with unknown pathogenicity, by medical professionals and researchers.

The use of this verification system ultimately offers the potential to deliver greater innovations in medical care for areas including treatment planning for genomic therapies for illnesses like cancer, accelerating the optimization of patient-centric medical care.