Research Report

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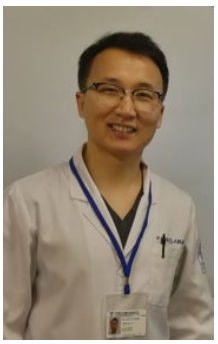
Research Period	From: 20200201 To: 20210131
Research Field	Molecular Biology



Research Theme at Tokai University

Title: QUALITY ASSURANCE OF NEXT GENERATION SEQUENCING (NGS)-BASED CANCER GENE PANEL TESTS

Abstract:

Next Generation Sequencing (NGS) is the technology that enables us to determine the sequence of DNA (deoxyribonucleic acid) with much longer footage in much shorter time than the conventional Sanger sequencing method. This technical breakthrough of NGS facilitates the application of the DNA sequence to various fields including the clinical applications of genome sequencing. People now have easier access to the clinical examination of genotyping of cancers, immune disorders, hereditary disorders, and so on, and this technology acquires more and more importance; the clinical use of NGS has been approved in the Japanese National Health Insurance. The accuracy of the assay in this application, however, is still yet to be evaluated before it gains the reliability by clinical practitioners and patients as much as the other standard laboratory tests have, and this is where we can contribute to the development of the technology.

NGS technology has long been applied to various fields such as basic molecular biology, environmentology, forensic medicine, natural history, agricultural science, and so on. Each of the fields has developed its unique ways to evaluate the reliability of the results, and the one in the clinical application has also been studied to some extent. The clinical application of NGS, however, is little studied to evaluate the reliability of the result with respect of the standardized medical practice. Even when it comes to NGS cancer panel test, the source of the samples, the reagents, the devices, the people who carry out the examination and the programs that analyze the data may vary from hospital to hospital. This research will be the first study to accumulate the result of NGS cancer panel test run by several hospital and commercial laboratories.

Enrolled in this study were a total of 7 laboratories which performed NGS oncology tests. The enrichment methods were variable among laboratories: amplicon-based and hybrid-capture sequencing methods used in 4 and 3, respectively. Two types of standards were prepared, i.e., genome-based (Thermo Fisher Scientific K.K) and DNA-based (Horizon Discovery Ltd.) samples and the variant allele frequency (VAF) of target genes was assigned. Those samples were shipped to the laboratories along with instructions for handling them and reporting the sequencing results. The samples were subjected to NGS tests in participant laboratories according to their protocols, then the result was reported.

Results / Achievements:

Please write a summary of your research results / achievements. Also, please share your research papers/articles, books, poster presentations if there is any.

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On-site evaluation as EQA for NGS oncology tests in the laboratory accreditation program under ISO 15189 was conducted as a pilot study. Quality indexes in the NGS result report from each of the participant laboratories were found acceptable. The types of the variant were categorized to SNP, MNP and INDEL, and the VAFs in the standards were confirmed within reference range provided by the manufacturers. The participant laboratories detected the targeted variants in the genome-based and DNA-based samples with success rates ranging between 86 to 100% and 75 to 100%, respectively, as compared with the reference ranges, and a few low-VAFs or false negative results were identified. Based on the feedback reports and self-assessment, auditors discussed with laboratory staffs for ongoing process and improvement in performance of the laboratory.
We have successfully implemented on-site evaluation as EQA for NGS oncology tests in the laboratory accreditation program under ISO 15189. The feedback reports in conjunction with self-assessment and discussion with auditors proved effective for evaluation and improvement of performance of the NGS oncology tests and the laboratory.□

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Any Additional Comments:

Please attach any photos of your research period, if there is any.