Journal of Medical Ultrasonics (2023) 50:271–272
https://doi.org/10.1007/s10396-023-01314-x

EDITOR'S NOTE

Is it possible to show a gene abnormality with an inherent macro morphology/function?

Yoshiki Hirooka[1]

Received: 16 April 2023 / Accepted: 21 April 2023

According to the latest statistics on trends in death rates (per 100,000 population) for leading causes of death in Japan, the death rate due to malignant neoplasms exceeded that of cerebrovascular diseases in 1981 to take the top spot, after which it has continued to increase while maintaining that top spot by an overwhelming margin. It is a scientifically proven fact that people are more likely to develop malignant neoplasms as they grow older, so this is no surprise given Japan's declining birthrate and growing proportion of elderly people. In addition, the death rate due to senility exceeded that of cerebrovascular diseases in 2018. The current leading causes of death in Japan are malignant neoplasms at number one, heart diseases excluding hypertension at number two, and senility at number three.

Let's refer to malignant neoplasms as cancer hereinafter. In terms of common treatments for cancer, the first choice in operable cases is complete resection so that there is no residual tumor. Anticancer chemotherapy is selected for unresectable tumors with local progression such as invasion of the major blood vessels, or in unresectable cases with distant metastasis. With respect to anticancer chemotherapy, a therapeutic agent (or combination of therapeutic agents)—which is selected based on clinical studies with a high evidence level such as randomized controlled trials—is regarded as the standard therapy for a certain cancer. However, standard therapy is a concept viewed from the perspective of "being effective in more patients". It turns a blind eye to the existence of a certain number of patients who do not respond to standard therapy. It is based on the idea of sacrificing the few to save the many.

Rapid progress has been made in precision/personalized medicine since the Precision Medicine Initiative was announced by then-President Obama during his State of the Union address in 2015. The Precision Medicine Initiative targets many disorders, not just cancer, but here I would like to talk a bit about cancer. The above-mentioned standard therapy and the therapy envisioned in the Precision Medicine Initiative are completely different. In the case of precision medicine, the focus is on mutant genes in cancer. Comprehensive genomic profiling (CGP) is performed using cancer tissue and blood, and if a therapeutic agent that targets the mutant gene causing the cancer is available, a therapeutic agent that is effective against the cancer is selected, in addition to standard therapy. We are currently in a transition period in which standard therapy and personalized therapy coexist, but we seem to be moving toward a future that will be dominated by personalized therapy.

The number of medical institutions where CGP can be performed is limited in Japan. CGP is available at only about 230 institutions in the whole country. Right now, many hospitals are forced to select standard therapy as first-line therapy despite the fact that some patients may not respond to standard therapy. They currently have no choice but to refer the patient to an institution that is capable of performing CGP only after the patient's condition has deteriorated with standard therapy.

If we can anticipate the presence of a mutant gene early on via a simple method, it will be possible to more rapidly refer the patient to an institution capable of performing CGP. In this month's issue, we have a groundbreaking report from Ikejima et al. about the ability to differentiate between *BRCA1* and *BRCA2* gene mutations based on clinicopathological and ultrasound characteristics in breast cancer patients. If we are able to judge the presence or absence of a particular mutant gene based on ultrasound imaging, it will allow us to anticipate a mutant gene early on via a simple method, making it possible for us to detect a gene abnormality using macro morphological/functional analysis. This means that medical ultrasonics has a part in personalized cancer therapy, and it is hoped that this field will continue to advance going forward.

✉ Yoshiki Hirooka
yoshiki.hirooka@fujita-hu.ac.jp

[1] Department of Gastroenterology and Hepatology, Fujita Health University, Toyoake, Japan

Data availability Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.