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Editorial

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Towards clinical implementation of circulating cell-free DNA in precision medicine

Wei Zhang[1,2]

[1]Department of Preventive Medicine, Northwestern University Feinberg School of Medicine, Chicago, IL 60611, USA.
[2]The Robert H. Lurie Comprehensive Cancer Center, Northwestern University Feinberg School of Medicine, Chicago, IL 60611, USA.

Correspondence to: Dr. Wei Zhang, Department of Preventive Medicine, Northwestern University Feinberg School of Medicine, 680 N. Lake Shore Dr., Suite 1400, Chicago, IL 60611, USA. E-mail: wei.zhang1@northwestern.edu

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Precision medicine holds the promise for precise diagnosis, prognosis, treatment response monitoring, and disease surveillance. Advancing clinical diagnosis is critical for providing precise and personalized care to patients and may improve clinical outcomes for many life-threatening diseases. Especially for cancers, which are a leading cause of death globally[1,2], early detection and precise diagnosis could provide opportunities to significantly reduce mortality as well as healthcare cost burden on the society[3]. For years, clinicians and researchers have been trying to discover better ways for more effective diagnosis and detection of cancers. At present, tissue biopsy pathology and imaging analyses are often the "gold standard" for cancer diagnosis and detection. Although extremely useful, tissue-based approaches are limited by various factors such as their invasive nature, inaccessibility to certain anatomical locations, and the issue of intra-tumoral heterogeneity, while conventional imaging-based approaches may be limited by possible exposure to radiation or lack of molecular information. For example, a particular biopsy procedure may miss the tumor or cannot capture the overall molecular features of the tumor, thus repeated biopsies are often required. Therefore, it is of urgent clinical need for the development of non-invasive or minimally-invasive approaches that can overcome those limitations associated with the current tissue- and imaging-based approaches.

During the past several years, assays based on patient-derived bodily fluids (i.e., liquid biopsy)[4], such as circulating cell-free DNA (cfDNA) in the peripheral blood, have demonstrated promising results as a clinically convenient, minimally-invasive approach for disease diagnosis, for example early detection of human cancers[5-9]. Circulating cfDNA, i.e., DNA fragments released by tumor cells or due to processes like apoptosis and necrosis, offers an intriguing alternative for cancer biomarker discovery, considering that cfDNA contains tumor-derived genetic and epigenetic information[7,10]. Notably, technological advances in genetic and epigenetic profiling have begun to allow interrogation of various molecular targets contained

in cfDNA for exploring their diagnostic potential[11-14]. Of particular interest are for example mutations and cytosine modifications (i.e., DNA methylation and hydroxymethylation) in cfDNA, although other features of cfDNA (e.g., fragmentation size) have also emerged to be useful clinical markers for diseases[15-17]. Though cancer biomarker discovery using cfDNA is expected to be probably the most active area of clinical implementation, cfDNA has also been shown to have diagnostic value for many other diseases or conditions, such as diabetic complications[18]. In this special issue "Application of circulating cfDNA in the diagnosis of human cancers and chronic diseases", we are interested in publishing research articles, short communications, technical notes, and review papers that cover the general topic of applying cfDNA in the diagnosis and detection of human cancers and other diseases. We look forward to discussing exciting progress and moving towards clinical implementation of circulating cfDNA in precision medicine.

DECLARATIONS

Authors' contributions

The author contributed solely to the article.

Availability of data and materials

Not applicable.

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Conflicts of interest

The author is a shareholder of Shanghai Epican Genetech Co. Ltd, which develops cancer biomarkers.

Ethical approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

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