
Preface

The high-throughput DNA sequencing method based on Sanger's principle provided an efficient tool for the sequencing of the whole human genome. With the help of capillary array platform invented by Professor Kambara, the human genome project was finished two years earlier than expected. In the post-genomic era, many biomarkers related to early disease diagnosis and personalized medicine have been found by the use of next-generation DNA sequencers. Although there are several DNA sequencing tools available for clinical detection of these biomarkers, pyrosequencing has an especially important role in identifying microbial genotypes, as well as in sequencing a short fragment containing the biomarkers related to personalized medicine.

Pyrosequencing is a sequencing-by-synthesis method based on the bioluminometric detection of by-products of inorganic pyrophosphates (PPi) during primer extension reaction. Although only several tens of bases can be sequenced during a run, it is enough for clinical routine use in detecting known biomarkers. As pyrosequencing is very suitable to the sequencing of short DNA sequence, the CFDA approved pyrosequencing technology for clinical use. Pyrosequencing is becoming the most popular DNA sequencing tool for personalized medicine in hospitals.

I have engaged in pyrosequencing research since 1999 when I came to Professor Kambara's research group in Central Research Laboratory of Hitachi. Professor Kambara directed me to perform pyrosequencing research in developing new chemistry for increasing the sensitivity, simplifying protocols for SNP typing, and miniaturizing the instrumentation. Professor Kambara, who is honored as an unsung hero of the human genome project by Science, is a highly reputed scientist in the DNA sequencing research field. I am very fortunate to have had his supervision in the research of DNA detection. Even after my return to China from Japan, he supported me in following pyrosequencing research for many years.

The aim of this book is to improve pyrosequencing protocols as well as instrumentation for better clinical use. Due to our continuous contributions, pyrosequencing is greatly improved in terms of template preparation, sensitivity, instrumentation, and detectable target species. We simplified the protocols of pyrosequencing by skipping the ssDNA preparation step. We enabled pyrosequencing to quantify the expression levels of mRNA and microRNA by coupling base barcodes into pyrosequencing. We adapted pyrosequencing to prenatal diagnosis of trisomy 21 by quantitatively pyrosequencing heterozygotes of fetal genomic DNA. We miniaturized the pyrosequencer by using a photodiode array as the light sensor. This book describes all of these improvements and novel applications of pyrosequencing technology.

There are 34 chapters in the book, and they are grouped as five parts: Part I is Advances in Pyrosequencing Template Preparation; Part II explores Pyrosequencing Technology Innovations; Part III delves into Multiplex Pyrosequencing based on barcodes; Part IV looks at Miniaturizing Pyrosequencing Equipment; and Part V examines Applications.

Now we are progressing toward precision medicine. To allow the right drug with the right dose at the right time, genotyping of an individual patient is required before prescribing a drug. I believe that pyrosequencing would be a useful tool in this progress. This book should be useful for people who are engaged in personalized medicine, disease control, and DNA diagnosis in other fields.

Professor Kambara, my teacher, retired from Hitachi in March 2015. I would like to dedicate this book to him, a great contributor to DNA sequencing technology.

Nanjing, China

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