Preface

The identification of sequence variation in DNA is a basic principle of genetic research. Numerous different methodologies have been developed over the past few decades, often focussed on a specific type of sequence change. The development of massively parallel sequencing approaches has made it financially and technically feasible for entire genomes to be sequenced in a rapid and cost-effective manner. Although this may seem to render many genotyping approaches obsolete, there are still a number of situations where specific, focussed assays are preferred. In this volume we have attempted to collate a broad range of different genotyping techniques.

Microsatellite analysis has many applications, including forensic identification and cell line verification. A description of a multiplex approach is provided in Chapter 1.

There may be occasions that specific sequence variants need to be genotyped. For a small number of variants in many DNA samples, High-Resolution Melt analysis (Chapter 2) and Taqman-based assays (Chapter 3) are attractive options. In situ analysis of variants in single RNA molecules is also possible (Chapter 4). For larger variant numbers, the MassARRAY system (Chapter 5) and Molecular Inversion Probes (Chapter 6) are powerful approaches.

Copy number variation (CNV) at diverse loci has been associated with a range of phenotypes, including disease. Accurate genotyping is problematic and may underlie contrasting reports in the literature. Different assays for accurately determining CNV are described here, including Pulsed Field Gel Electrophoresis (PFGE, Chapter 7), Paralogue Ratio Test (PRT, Chapter 8), Multiplex Ligation-dependent Probe Amplification (MLPA, Chapter 9), Emulsion Haplotype Fusion PCR (Chapter 10), and Droplet Digital PCR (ddPCR, Chapter 11).

In many cases a genotype alone is not sufficient information; it is also important to know on which alleles each variant is located. For combined genotyping and haplotype generation of large stretches of DNA, there are different NGS-based approaches: long range PCR combined with PacBio sequencing (Chapter 12) and Targeted Locus Amplification (TLA, Chapter 13).

Although most techniques can be applied to DNA from almost any source, some assays have been specifically optimized for certain types of organism. For bacteria, Multilocus Sequence Typing (Chapter 14) and Rapid SNP detection with pyrosequencing (Chapter 15) are described. Genotyping-by-sequencing for plant analysis is also included (Chapter 16).

Last, but certainly not least, it is critical for genotyping findings to be reported in a clear and unambiguous fashion. A summary of the most pertinent points when describing genetic variation is included (Chapter 17).

Leiden, The Netherlands — *Stefan J. White*
Seattle, WA, USA — *Stuart Cantsilieris*

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