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## Preface

### “Your genome is an email attachment”

What a difference a few years can make? In 2001, to a global fanfare, the completion of the first draft sequence of the human genome was announced. This had been a Herculean effort, involving thousands of researchers and millions of dollars. Today, a project to re-sequence 1,000 genomes is well underway, and within a year or two, your own “personal genome” is likely to be available for a few thousand pounds, a price that will undoubtedly decrease further. We are fast approaching the day when your genome will be available as an email attachment (about 4 Mb). The key to this feat is the fact that any two human genomes are more than 99% identical, so rather than representing every base, there is really only a requirement to store the 1% of variable sequence judged against a common reference genome. This brings us directly to the focus of this edition of *Methods in Molecular Biology*, Genetic Variation.

The human genome was once the focus of biology, but now individual genome variation is taking the center stage. This new focus on individual variation ultimately democratizes biology, offering individuals insight into their own phenotype. But these advances also raise huge concerns of data misuse, misinterpretation, and misunderstanding. The immediacy of individual genomes also serves to highlight our relative ignorance of human genetic variation, underlining the need for more studies of the nature and impact of genetic variation on human phenotypes.

In March 2009, the US Congress passed the American Recovery and Reinvestment Act, which, among other things, granted the US National Institutes of Health an additional \$8.2 billion in funding to disburse over the next 2 years. A substantial amount of this investment is likely to be channelled towards the re-sequencing of thousands of additional human genomes. When combined with the substantial amounts of data that already exist, data availability will no longer be a barrier to the understanding of human genetic variation. Against this background, we feel this edition of *Methods in Molecular Biology* is probably very timely. Although no publication could hope to comprehensively address all forms of human variation, our contributors have tried to provide coverage of most forms. This includes single nucleotide polymorphisms (SNPs), insertions/deletion (indels), copy number variation (CNVs), variable number tandem repeats (VNTRs), mitochondrial variation, mobile elements, and epigenetic variation. In the tradition of the series, we consider both laboratory and in silico methods, in many cases both in the same review. We believe that this underscores the need for increasing interactions between bench scientists and bioinformaticians. Neither breed of scientists can be independently successful in understanding the full impact of variation, but by working together they may have a fighting chance.

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