Although the term “junk” DNA was used since the early 1960s, the term’s origin was attributed to Susumo Ohno who officially used the term to describe pseudogenes’ sequences resulted from gene duplication and subsequent mutagenesis events. Since then, the term was widely used to describe any non-coding sequence of the genome. Today, “junk” DNA refers to any genomic sequence that does not play a functional role in the organism. The use of the term was accompanied by various unanswered questions: Why do we have so much “junk” DNA in our genome? Do these non-coding sequences have functional significance? The discovery of novel genomic elements in the recent years was a step forward in an attempt to address these issues. It appears that the percentage of the non-functional DNA is being significantly reduced as more and more functions are attributed to those non-coding regions of the genome. Despite the continuous shrinkage of the non-functional portion of the genome, it is believed that a significant part of the genome is indeed non-functional.

In this book, we attempt to provide a thorough review of various non-coding genomic elements and discuss in depth their role in health, disease and evolution. We begin our exploration with non-coding RNA molecules, miRNAs, piRNAs, LncRNAs and transposable elements as these moieties dominate the scientific literature in the last 10 years. We proceed with the discussion of copy number variation regions, mini- and micro-satellites, and proximal and distal elements of the genome. The last section of this book focuses on the review of well-known non-coding regions of the genome, introns, centromeres and telomeres, but enriched with newly discovered functions. As the vast amount of data in regard to these elements is attributed to a great degree to the growing technology in the field of biomedicine, the last chapter of this book discusses the latest development in the field of Next Generation Sequence and the potential applications of this technology in the study of non-coding regions of the genome.

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