Preface

The DNA double helix, as we know it since the seminal paper of Watson and Crick in 1953 (Watson, J.D. and Crick, F.H.C. A structure for deoxyribose nucleic acid. *Nature* 171: 737–738, 1953), probably a special variant of a primordial RNA, has been (and still is) under heavy environmental pressure. It is exposed to chemical and physical agents such as drugs and radiation that altogether have altered nucleic acids since their existence. Therefore, DNA is subject to continuous changes in base composition and structure, generally called mutations (Latin *mutare*: to change). These changes not only shaped the DNA molecule, but eliminated or introduced new information, which was, is, and will be fundamental for evolution. Certainly, evolution would not have been possible without mutation(s). The immense phenotypic variations, which surprise the alert observer, rest upon genetic variation (i.e., mutations). Although large subsets of mutations are beneficial for the survival and evolution of a species, certain mutations can have a detrimental impact on individuals – they may indeed lead to a reduced fitness and diseases down to death. These ambivalent aspects of mutations shape life on Earth.

Mutations range in size from single base exchanges, single base deletions, small deletions, inversions, insertions, and duplications of two or a couple of bases more, to large changes of several kilobases to megabases and chromosome aberrations such as supernumerary chromosomes, missing chromosomes, translocated parts from one chromosome to another one (or others, and vice versa), and aberrant physical appearance of chromosomes (e.g., in various forms of leukemia or in the case of ring chromosomes). This listing is by no means a complete description of all possible mutations, but outlines the variety of potential changes at the molecular and chromosomal level. Keeping in mind that before the discovery of DNA by the pioneer Friedrich Miescher around 1869 in the nuclei of leukocytes from pus on surgical bandages (at that time coined nuclein), mutations were already known in various organisms, but could, of course, not be related to DNA. For example, albinos in plants, animals, and humans were considered exotic and unexplainable erratic forms of life.

Today we know of many chromosomal, genomic, and genic mutations, and as more and more genomes are sequenced and resequenced, more and more muta-

The Handbook of Plant Mutation Screening. Edited by Günter Kahl and Khalid Meksem

ISBN: 978-3-527-32604-4

tions are discovered and will serve to identify genetic differences between individuals or species, or the causes underlying the 6000–8000 inherited disorders in humans. It is for this reason that the detection of mutations became an important discipline in biology and medicine, and therefore the techniques of discovery and diagnostics of these changes are rapidly evolving.

This book witnesses the importance of mutations. The detailed description of mutation screening technologies by renowned experts in the field gives a flavor of the past and present state of mutation sciences, but also a sense of what is emerging in the area of biophysics, biocomputing, molecular biology, and genomics.

Carbondale (USA)
Frankfurt am Main (Germany)
December 2009

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