Preface

'An organism's astonishing gift of concentrating a 'stream of order' on itself and thus escaping the decay into atomic chaos – of 'drinking orderliness' from a suitable environment – seems to be connected with the presence of the 'aperiodic solids', the chromosome molecules, which doubtless represent the highest degree of well-ordered atomic association we know of – much higher than the ordinary periodic crystal – in virtue of the individual role every atom and every radical is playing here'.

E. Schrödinger, 1944. What is Life? Cambridge at the University Press.

'We feel that the patterning of chromosomes is of far greater importance than hitherto realized, yet we have not yet found the tools permitting us to analyze this phenomenon'.

E. Mayr, 1959. Where are we? Cold Spring Harbor Symp. Quant. Biol. 24.

'A logical analysis of the concept of organism leads us to look for organising relations at all the levels, higher and lower, coarse and fine, of the living structure'.

J. Needham, 1950. Biochemistry and Morphogenesis, Cambridge University Press.

'However, as long as the chemical nature of the genetic material remained unknown, genetics had to remain a rather abstract branch of the biological sciences'.

W. Arber, 1979. Promotion and limitation of genetic exchange, Science 205, 361–365.

This is a book about order. It treats the order present in molecules and chromosomes.

Physical particles, atoms and molecules are rigidly organized structures in which polarity and symmetry prevail. The aim of molecular cytogenetics is to find out how molecular order determines chromosome order and how in turn chromosome order

has contributed to shape the order found in heredity, embryological development and evolution.

Order in living organisms and cellular components is both obvious and elusive. It is obvious because all of us recognize at first sight the bilateral symmetry of the human body or the polarity in the growth of a tree. In the cell the regular division of the chromosomes, or the maintainance of the same pattern in the different organelles throughout cell generations, are manifestations of order which are easy to grasp. But how superficial is this order, or better formulated, how deep does it go? What is actually elusive is the degree and the interdependence of order.

How is the symmetry of the organism related to the symmetry of the chromosome and this in turn to the symmetry of DNA, and this in its turn to the symmetry of the carbon atom? It is here that the task begins, and it is a formidable one, since there are no obvious links, nor even guidelines to help us in the search for these relationships. The reason is simple. The amount of ignorance is impressive, especially at the molecular and cellular levels.

Heredity, embryological development and evolution are extremely ordered processes. If they were not so they simply could not be recognized or characterized. Heredity is the expression of an ordered process in the transmission of potentialities to the progeny. This has allowed the formulation of this transmission in the form of 'laws'. Nothing could be more ordered than the development of a fertilized egg into an organism. The smallest deviation or error from the programmed pathways leads to irreparable deformation or death. Evolution in itself is the recognition of order and hierarchy in the formation of organisms. Instead of their separate and chaotic creation on the earth, they are all linked by having descended from previously existing organisms in a definite time sequence. Moreover, evolution developed in a hierarchic way. The simplest organisms appeared first, the more complex in later times.

Our main concern is the order within the chromosome. How it came about and what consequences it had. But since our ignorance of this organelle is so profound one needs to find first of all whether the chromosome shows any organization, what kind of organization it has, and what mechanisms are responsible for this organization. My main task has been an attempt to assemble a body of information which has been available for the last 30 years, and which has accumulated rapidly in recent times, but due to the fact that it has been left dispersed, it did not create a coherent picture and as such did not make order evident. I have not tried to see order. It cannot simply be seen at a glance, or by inspecting the data available. I have painstakingly collected the data which I found scattered, to see to what extent they could give us a picture either of disorder or of order. To my surprise there seems to be much more information which points to order than one would expect.

Once put into a coherent scheme many chromosomal phenomena which before seemed to point only to disorder show the reverse. Moreover, as the analysis deepens, due to a more extensive study of a phenomenon, or by analyzing it at the molecular level, the processes which before appeared as isolated cases or as disconnected events appear now bearing definite relationships to others and are found to occur throughout most species or phyla. However, it would be premature to expect to be able at present to acquire an all-embracing view of most phenomena related to

chromosome organization and evolution. But guidelines for experimentation and new principles emerge naturally from assembling the previously dispersed molecular and structural information.

The picture that emerges at this stage is one that will be revised many times in the future through adjustments and startling refinements. What can be put together now may only serve as the raw guidelines for the explosive growth of research occurring in the field of the molecular analysis of the eukaryotic chromosome. To write any book today on chromosomes with the intent to clarify their organization is a severe experience due to the impressive flow of literature. For this reason I had to rely many times on review articles. However, the book is mainly based on over 25 000 reprints from my collection supplemented by about 3000 photocopies. There has been no intention to cover the literature exhaustively but only to assemble relevant data.