

nature genetics

volume 1 no.1 april 1992

Nature creates Nature Genetics

Almost exactly 39 years ago (on 25 April 1953), J. D. Watson and F. H. C. Crick observed, at the end of their account of the structure of DNA, that "It has not escaped our notice that the specific pairing we have postulated [between purine and pyrimidine bases] immediately suggests a possible copying mechanism for the genetic material" (*Nature* 171, 737; 1953). That now-famous throw-away line was the beginning of molecular genetics, which has transformed the classical concept of a gene into something intellectually tangible — a string of nucleotides in a molecule of DNA. Those whose investigations of genetic structure now prosper because of this development should occasionally pause to marvel at how much classical geneticists were able to accomplish while still ignorant of what genes are.

And everybody should marvel at what has become of molecular genetics. Strictly speaking, it should not be surprising that many hereditary diseases have been traced to the simple substitution of a single nucleotide by another (as in the inheritance of many forms of aberrant haemoglobin, including the one responsible for sickle-cell anaemia, for example), yet the now-long list of diseases whose origin is as simple is clear proof of how literal the link often is between phenotype and nucleotide sequence.

Yet there have been surprises, notably the discovery that some inherited diseases arise from the uncontrolled replication of particular trinucleotide codons at unstable regions of the genome: the recent excitement about the genetics of myotonic dystrophy (*Nature* 6 February 1, 545–551; 1992) is vivid proof of how much remains to be learned about the link

between genome structure and disease. (An article in this issue may be the first identification of an inheritable disease caused by aberration in the gene controlling the production of a regulatory element, page 56.) This will be an important focus of the attention of *Nature Genetics* in the years ahead.

Researchers should not be dismayed that developments like these are widely reported in the general press. That is merely a measure of the widespread compassionate interest in inheritable disease. Who can be but flattered by such public testimony to the importance of a field of research?

The research community's interest, rather, is that there should also be a wide general understanding that the identification of an aberrant gene does not imply that there is a cure at hand for the condition for which it is responsible. After all, the discovery of the mutation of the *ras* gene linked with the occurrence of solid tumours in people is now a decade old, but who can yet say that the mechanism of the link with cancer is understood? The elucidation of the mechanisms by which genes determine the behaviour of the cells that carry them will be a general preoccupation in the years ahead. *Nature Genetics* intends to play its part in the publication of this important research, and also of course, in classical genetics that throws light on the human genome.

Much will be learned about the mechanisms by which human genes function from studies in other than human organisms. One of the striking lessons of the past 39 years is the extent to which all organisms depend on common biochemical mechanisms, themselves controlled by genes that are very similar to human genes. Much will be learned about the

human genome from the similarities to and differences from the genomes of other organisms, perhaps especially, the simple nematode *C. elegans*. Willingly, *Nature Genetics* offers itself as a vehicle for the publication of all research that promises to illuminate human genetics.

Techniques are also important. Indeed, one of the most remarkable features of the brief history of molecular genetics, and of molecular biology in general, is the breathtaking interplay between technique and discovery. Ever since the discovery of reverse transcriptase independently by David Baltimore and Howard Temin in 1971 (*Nature* 226, 1209, 1211; 1970), it seems that successively more sophisticated demands for information about the structure of genes and their placement in the genome have been met by increasingly sophisticated techniques, the development of the polymerase chain reaction (PCR) for making copies of single DNA molecules for example. *Nature Genetics* will consider for publication accounts of technical innovations with a bearing on the understanding of the human genome.

Ambitions to determine the entire nucleotide sequence of the human genome, represented in the United States by the Human Genome Project, will be a powerful incentive for further technical innovation. The article in this issue on page 34 is an interesting illustration — a demonstration of technical innovations to determine automatically the nucleotide sequence of a 106 kilobase-long stretch of DNA. Few will be surprised that the power of the technique appears to be limited by the sophistication of the algorithm used to match fragments of the DNA. *Nature Genetics* will be interested to hear from contributors who can improve on the software used in this and other contexts.

Two other foci of interest deserve attention, one of which is readily identified by the general attention it has been given in recent years: the application of knowledge of the structure of genes in fields as different as the diagnosis of disease and the use of nucleotide sequences in forensic investigations. These are powerful techniques whose use has already prompted much public argument, even anxiety. The prospects of gene therapy, which lie mostly in the future, are technically of absorbing interest, offer remarkable opportunities for medicine, but also raise important social issues. *Nature Genetics* will publish research promising important developments in these fields, and will also aim to keep its readers abreast of the ethical arguments surrounding them.

Finally, there is the bearing of molecular genetics on the understanding of general problems in biology. In spite of the attention given to the mechanism of speciation by the generations of biologists since Darwin, important issues remain unresolved. So, too, do particular questions about the evolution of *Homo sapiens*. Genetics as now practised offers the prospect of answering some of these grand questions. Essays in this direction will also be grist for the mill of *Nature Genetics*.

The importance of all these developments is self-evident and generally accepted. But if a research field is important, do not the discoveries that emerge from it deserve to be published quickly? That is another of the guiding principles of *Nature Genetics*. The overall objective is to provide professional researchers in the field with a vehicle for the publication of important research, and to provide a wider readership with a means of comprehending a field of research that is at once quickly moving, of great importance for scientific understanding and which promises powerfully to influence the development of society in the years ahead.

But are not these also the objectives of *Nature*? Of course. The obvious difficulty is that, with the research material arising from such an active field, even a weekly journal cannot accommodate the flood of what needs to be published quickly. *Nature Genetics* owes its existence to *Nature's* conviction that another vehicle for the publication of research in genetics would serve the public interest. It is not intended that *Nature* should publish less genetics, but that the two journals together should publish a great deal more.

How will this work? *Nature Genetics* is at once a part of *Nature* and separate from it. Within the general framework of an agreed editorial policy (which includes the same criteria of excellence for both journals), *Nature Genetics* will benefit from less restrictive rules on the allowed length of manuscripts. But would-be contributors must make their own guesses about the relative advantages of sending their contributions to one journal or the other.

The day-to-day link between the two journals is that they share certain facilities and that there may be occasions when *Nature* offers to contributors whose manuscripts it cannot publish for lack of space that it will, if asked, pass on their manuscripts and the accompanying referees' reports to its sister journal. *Nature* will also give its general readership an account of what *Nature Genetics* has just published. The overall objective is more faithfully to reflect the interest of this important field.