Annual Review of Genomics and Human Genetics, 2009. Aravinda Chakravarti and Eric Green (eds). Annual Reviews, 4139 El Camino Way, P.O. Box 10139, Palo Alto, CA 94303-0139, USA. Vol. 10. ix + 551 pp. Price: US$ 84.

Those interested or actively involved in the field of genetics and genomics will find a number of their favourite topics covered in the 23 chapters of this volume. The book provides a review of recent advancements in genomics and the methods that can be used to study mechanisms involved in disease, health or evolution of genomes and genomic landscapes.

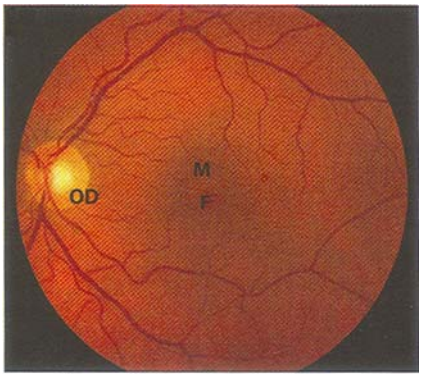
Diverse aspects of host genetics and host–pathogen genomics as well as methods to dissect their genetic basis and elucidate mechanisms of pathogenesis have been dealt with comprehensively in chapters 2, 4, 5, 11 and 19. Chapter 2 provides an account of how integration of leads from genetic, biochemical and genomic studies with the molecular process of ageing has provided insights into the molecular pathogenesis of age-related macular degeneration. Genetic dissection of chronic pancreatitis (CP; chapter 4) has reaffirmed the century-old hypothesis that it is an autodigestive disease and the genes found to be associated contribute to different pathways in trypsinogen activation and different etiology of CP. Chapter 5 reviews the 30 genes that have been associated with Crohn's disease and how these could be involved in dysregulation of mucosal immune response against the luminal flora. Chapter 19 deals with studies on identification of genes and variations popularly called performance enhancing polymorphisms (PEPs) that affect the outcome of athletic performance. Nearly 200 genes affecting a variety of functions have been documented to affect athletic performance. A downside to identification of such targets is the growing concern that practice of gene doping is likely to increase.

Chapter 11, one of the most illustrative and best chapters, demonstrates how genomics methods have provided a new perspective on schistosoma biology and helped dissect various aspects of host–pathogen genetics. Schistosoma has a complex life cycle that includes free-living aquatic stages in addition to parasitic stages in the intermediate host snail and in the definitive mammalian host. Functional genomics analysis across genders and developmental stages within mammalian and snail hosts have provided a framework for developing new antischistosome vaccines as well as drug targets and diagnostic markers for control and treatment of schistosomiasis. This chapter demonstrates beautifully the relevance of a basic education in zoology, and a revision of its curricula in undergraduate education is likely to attract a large number of young researchers to this field.

The book also deals with various aspects that contribute to genomic architecture and impacts disease phenotypes. These relate to variability in chromosomes, the telomeres, copy number variations, post-translational modifications and GC-biased gene conversion events (chapters 1, 3, 8 and 21). In the first chapter, Janet Rowley has uniquely chronicled the developments in clinical cytogenetics through her account of the workshops which she has held in the field right from its days of inception. These workshops have enabled scientists globally to compare different chromosomal breakpoints in diseases, follow-up of survivors, allowed sharing of reagents and results, and also eventual cloning of the causal genes. Chapter 3 deals with syndromes related to shortening of telomeres, a phenomenon that has been described only in recent times. A large number of phenotypes linked to premature ageing, features like nail ridging, idiopathic pulmonary fibrosis, liver fibrosis, decreasing bone marrow cellularity and function, thrombocytopenia, immune dysfunction, increased cancer risk, chemotherapy and radiation therapy intolerance have been linked to telomere shortening. Chapter 8 deals with yet another aspect of defects in the molecular machinery related to post-translational processing of prelamin A that has been implicated in progeroid syndromes.

The often neglected phenomenon of gene conversion, wherein short-scale unidirectional changes accompany recombination events, has been dealt with in chapter 14. GC-biased gene conversion events have been reported in many eukaryotes and appear similar to selection. The authors have discussed its functional consequences and also sounded a cautionary note to the evolutionary biologists for not neglecting this aspect in neutral models of sequence evolution for detecting selection in genomes. Another important feature of the genome dealt with in chapter 21 is copy number variation (CNV), which through various molecular mechanisms like gene disruption, dosage effect, position effect, gene fusion, etc. has been implicated in phenotypic diversity. The challenges in understanding CNV genotype-to-phenotype correlations have been dealt with in great detail. Heritable variation in gene expression constitutes an important source of observed phenotypic variability within and between populations. Chapter 15 reviews how genome-scale dissection of transcriptional variation has led to the identification of gene expression, quantitative trait locus, and provided detailed insights into the complex genetic architecture of transcriptional variation.

Various aspects of evolution ranging from evolution of primates and sex chromosomes to genomic mechanisms like imprinting and recruitment of proteins into animal venoms have been dealt in chapters 12, 14, 16, 17 and 22. Chapter 12 reviews the insights on the origin and evolution of imprinting mechanisms in mammals, especially the role of repetitive element through analysis of imprinted gene clusters in marsupials and monotremes. Chapter 16 provides an account of how comparative genomics studies have revealed that distant species may share fundamental properties of sex chromosome evolution, whereas very similar species can evolve unique sex chromosome systems. Chapter 17 deals with analysis of completed sequences of various primate genomes that have provided a framework for understanding primate genome evolution. Gene regulation and expression as opposed to amino acid replacements seems to have driven



Fundus photograph showing the retina of a normal individual. Retinal blood vessels are clearly visible. F, Fovea; M, Macula, and OD, Optic disc.

primate adaptations to new environments. The authors have emphasized the need for striking a balance for quality and completeness of the primate genomes versus more number of genomes, and also the challenges in functional elucidation due to the paucity of adequate material either because of sample accessibility or their dwindling numbers.

The book has also devoted chapters (6, 7, 13, 15 and 18) that describe the essential tool kit that a genomic scientist needs to be aware about. Contemporary genotyping technologies as well as the anticipated future developments using microfluidics and nanotechnology and use of next-generation sequencing platforms for transcriptome and genome analysis is likely to enable studies at much higher throughputs, finer resolution and lower costs. Transcriptome sequencing (described in chapter 7) has enabled genome annotation, alternate isoform discovery, gene expression profiling, mutation profiling, non-coding RNA discovery and detection, identification of aberrant transcriptional events and discovery of RNA editing sites. With the leads obtained from GWAS studies and the increasing emphasis on the role of rare variants in diseases, methods are also being developed to capture/enrich genomic regions for targeted re-sequencing in thousands of individuals. Issues relevant to genomic partitioning have been dealt with extensively in chapter 13. Chapter 18 discusses how genotype imputation has enabled integration and meta analyses of GWAS studies conducted on different array platforms from different studies, increased the power of genome-wide scans, and facilitated and also led to the discovery of causal single nucleotide polymorphism from reference sequences after association studies. With the availability of complete sequences from 1000 genomes project (www.1000genomes.org) and reference databases in diverse populations, genotype imputation would be a key tool in genetic studies.

A few chapters deal with the impact of genomics on common man in terms of testing its impact. It also deals with the uncomfortable domain of ethics in genomics research where sometimes the quest for answers supersedes the responsibility and accountability in handling biospecimens (chapters 9, 10, 20 and 23). These could be important issues which we might need to evolve for setting up national guidelines. Chapter 9 has provided an overview of how genetic testing has become quite widespread and successful in Israel due to the efforts of the socialized medical system, the government national programme for the detection and prevention of birth defects, a central registry of genetic disorders and the availability of a medical genetics unit. Simultaneously, legislation to regulate genetic testing and protect privacy to avoid discrimination has also been implemented. There is also a concern of the custodianship of and access to DNA specimens, and attached clinical and genetic data that are held in biobanks and large disease cohort collections. It is being realized that an exercise in de-identifying or anonymizing the large GWAS dataset is futile, and a need is being felt to devise comprehensive guidelines that not only allow scientific advancements but also respect the patient's confidentiality and rights while protecting the investigators from legal challenges. Chapter 10 has described a few salient points that need to be considered, discussed and implemented in this regard. Chapter 20 deals with issues of direct-to-consumer testing where many companies are now providing genetic tests without recommendations of medical experts. This is especially important for mutations with low penetrance which cause disease in only a minority of patients and could lead to a large number of false positives. Low penetrance genes pose a risk in population-based screening, since by definition many persons who test positive for low penetrance variations might never develop the disease, but this could influence reproductive choice, health behaviour and potential for discrimination on the part of the insurance company or the employer.

The last chapter provides an account of genetic studies that have been carried out over the last 40 years in the Amish and Mennonite populations of North America, represented by a limited number of individuals in the early eighteenth century. This chapter provides a historical account of genetic studies conducted in this population that led to mapping mutations and founders for many diseases, and also the efforts by researchers and companies in setting up facilities in this population, which have not only been useful in rapid genetic screening but also in administering timely dietary interventions to change the course of diseases. Some of the primary objectives of the Human Genome Project – to harness genetic knowledge to heal the sick, prevent disability and reduce medical costs, have been realized through an integration of genetic technologies into a rural pediatric practice. Studies in these populations have also highlighted how discovery in a limited sample size could be applicable to larger populations. The chapter also deals with an important aspect of how for effective community participation for a genetic study, it is imperative that scientists must first commit to caring for individuals.

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Worldviews – An Introduction to the History and Philosophy of Science (Second Edition). Richard DeWitt. Wiley–Blackwell, West Sussex, UK. 2010. 376 pp. Price: AU$ 39.95.

As an academic teaching the subjects 'Research methods' (first year Master's students of sustainable agriculture) and 'Research philosophies and methods' (fourth year research Honours students of agriculture), I enjoy reading new books and new editions of books dealing with themes on the history and philosophy of science (HPS). My enjoyment is because such readings enable me to teach my students better by raising provocative questions and providing new problems to set a context wider than they usually perceive and experience. I was, therefore, happy to get a copy of the new edition of DeWitt book.

Before I go into discussing the book, I would like to mention that a sound understanding of the HPS is critical in the context of Indian high school-, college- and university-science teaching. Bulk of high school-, college- and university-mainstream science teaching in the West includes HPS, at least for background understanding. For example, the curricula include either cultural information or