

Annual Review of Genomics and Human Genetics, 2010. Aravinda Chakravarti and Eric Green (eds). Annual Reviews, 4139 El Camino Way, P.O. Box 10139, Palo Alto, CA 94303-0139, USA. Vol. 11. 461 pp. Price: US\$ 84.

The book under review is a collection of interesting articles covering various aspects of genetics and genomics. Broadly, the book covers the areas of basic genetics, medical genetics, evolutionary genetics, and ethical, social and legal aspects of human genetics, giving the reader a flavour of recent advances in these fields.

The book begins with an article on genomics of long-range regulators of gene expression. Transcriptional regulation of gene expression by locally acting elements like promoters and transcription factors has been studied in detail for a large number of genes; however, there is little information available on long-range controllers of gene expression like enhancers, suppressors, etc. The article reviews various known long-range regulators, with appropriate examples, and further dwells on the various genome-wide approaches to identify these long-range regulators. The mitochondrial genome which was sequenced three decades ago is now known to encode about 1000 proteins by taking advantage of recent advances in proteome analysis. Details of mitochondrial proteome and the methods used to study it form the subject of the second review in this book (p. 25). The authors argue that once all the proteins of the mitochondria are characterized along with their interactions, systems biology approaches could be applied to study them, as has been done for smaller genomes like *Escherichia coli*, for better understanding of mitochondrial functions.

Population genetics studies have relied on linkage maps of human genome obtained from various markers like microsatellites to the more recent genome-wide single nucleotide polymorphism maps. Two articles in this volume review various aspects of linkage maps and their utility in genetic research. The article on quantification of fine structure of human recombination (p. 45) compares the linkage disequilibrium maps derived from pedigree-based human recombination data with those obtained from sperm genotyping methods. The authors conclude that both these maps

generally agree at gross levels, but there are significant differences between them at local levels. The article on admixture mapping (p. 65) traces the history of admixture mapping strategy and its utility in various genome-wide association studies in various multifactorial disorders like hypertension, multiple sclerosis, diabetes, etc., especially in the African-American population.

This book has generously devoted five articles to the genetics of human diseases. The article on genetics of hair development and diseases associated with abnormalities of hair provides a detailed overview of the various genetic diseases wherein hair abnormality is one of the phenotypic features. It is a must read for researchers in this field. Many of these diseases are also associated with early onset cognitive impairment and this forms the topic of the next review on genetics of mental retardation (p. 161). The author has reviewed in detail, various genetic factors associated with developmental delay, including novel microdeletion syndromes detected by recent array comparative genomic hybridization approaches and X-linked forms of mental retardation. It will be an interesting read for medical geneticists and physicians involved in care of children with mental retardation. Skeletal dysplasias is a group of disorders associated with abnormality of bones and the recent advances in genetics of these diseases have revealed a wealth of information regarding the number of signalling pathways which are perturbed in these diseases. The review of these signalling pathways and diseases resulting at each level in these pathways is an informative and educative read for researchers in this field (p. 189). The authors have provided a lucid description of various signalling pathways using excellent diagrams. Another important disease caused due to perturbation of signalling pathways is cancer. The review on cancer genomics (p. 133) highlights the approaches used to study the cancer genome and further dwells on application of molecular profiling of cancer in decisions regarding the treatment of malignancies, using breast cancer as a prototype.

Genetics of coronary artery disease has been reviewed using a summary of various genome-wide association studies which have provided with a number of risk loci for myocardial infarction (p. 91). However, these risk factors

account for only a minimal increase in the odds of developing the disease and hence their causality is difficult to establish. In spite of this, these risk alleles are being used by companies/laboratories to inform consumers about their risk of developing these diseases. The last article in this book deals with the important issue of direct to consumer genetic testing (p. 427). A number of companies have been set up in USA and Europe in order to offer genetic tests directly to consumers/people without the involvement of a geneticist. This has led to heated debates in various forums regarding the dangers of such an approach. The authors discuss these issues using a representative research case study (Multiplex Initiative).

The book presents a flavour of evolutionary genetics through a bunch of five articles devoted to this area. The review on evolution of lactation in mammals (p. 219) supported by genomic data across various species provides an overview of the origin of lactation and molecular evolution of lactation. The next article discusses the evolution of reptiles at chromosomal and single-gene levels (p. 239), including the recent sequencing of *Anolis* lizard genome and various genome resources available for the study of nonavian reptiles. The article on neutral theory of evolution (p. 265) critically reviews the various statistical methods, beginning with Bayesian methods to the more recent theoretical studies and genome-wide analyses of natural selection. Since the publication of Charles Darwin's *On the Origin of Species*, the process of speciation has remained an enigma. The review on chromosomal speciation (p. 291) aptly examines the classical and current models of chromosomal speciation and will serve as a good reference for students of evolutionary genetics. The fifth article in the series gives an interesting overview of the conflicts between the theories of 'creationism' and evolution (p. 317). The authors have termed these as 'evolution wars' and have provided a graphic description of ongoing challenges facing the education of evolution, right from the level of the classroom to the legislature.

Four articles in the book deal with the emerging social, ethical and legal issues in genetics. Issues pertaining to public attitudes and perception of genetics on topics ranging from genetic testing to genetically modified foods have been

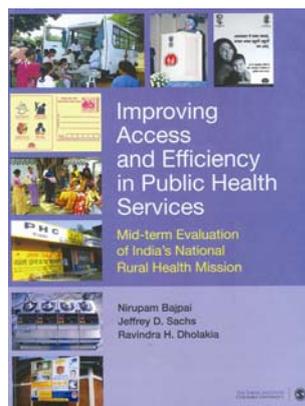
BOOK REVIEWS

dealt with in detail by the authors (p. 339). Human genetics is moving swiftly from the research laboratory to its application in human diagnostic testing as well as therapy. This has led to the emergence of a number of issues relating to safety, confidentiality and genetic discrimination of human subjects. The article on informed consent in genetic research (p. 361) lucidly brings out the paramount importance of informed consent in human genetics testing and research. The authors present a detailed description of the steps involved in obtaining informed consent. The article will be useful for researchers embarking on human genetics research. Inventions and innovations resulting from research are an important intellectual property of the researcher/institution, and need to be protected from unlawful commercialization. The article on patents (p. 383) dwells on various aspects of patents, right from the definition of a patent to examples of various instances of patent infringement in great detail. The authors have presented famous examples like erythropoietin and polymerase chain reaction patents to emphasize the importance of patenting the innovations.

Overall, the editors have put together an educative collection of articles dealing with varied aspects of human genetics and genomics, ranging from evolutionary genetics to contemporary issues, which will be an interesting read for researchers in this field.

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Improving Access and Efficiency in Public Health Services: Mid-term Evaluation of India's National Rural Health Mission. Nirupam Bajpai, Jeffrey D. Sachs and Ravindra H. Dholakia. SAGE Publications India Pvt Ltd, B-1/I-1, Mohan Cooperative Industrial Area, Mathura Road, New Delhi 110 044, India and The Earth Institute, Columbia University, 405 Low Library, MC 4335, 535 West 116th Street, New York 10027, USA. 2010. xviii + 117 pp. Price: Rs 695.

The Indian Government spends less than 1% of the GDP on health. The National Rural Health Mission (NRHM), spanning a period of seven years, from 2005–06 to 2011–12, began in April 2005 with the aim of improving public health services while focusing on states having a weak public health infrastructure. NRHM is a Central Government initiative that intends to bridge the north–south divide in healthcare facilities. One of the elements of NRHM is the commitment of the Central Government to increase public spending on health from 0.9% to 2–3% of GDP. Mainstreaming of Ayurveda, Yoga, Unani, Siddha and Homeopathy manpower is one of the supplementary strategies of NRHM for achieving the set goals.

The book under review is a report on the mid-term evaluation of the functioning of NRHM. The health sector in the rural India has been criticized for its inefficiency and the low quality of services, particularly in the northern and central parts of the country. The major sufferers of poor health amenities are eventually the rural women and children. Under the study, field work was carried out in five districts from Uttar Pradesh, Madhya Pradesh and Rajasthan. Accredited Social Health Activists (ASHAs), who provide basic curative medicines, first aid and preventive care, and participate in healthcare delivery programmes, were the main focus of the study. A trained ASHA is considered to be the most critical element of NRHM.

Indeed, NRHM has helped increase the rate of fully immunized children (except in a few states). It has also helped reduce the infant mortality rate in some states, but the study indicates that this rate is clearly not even close to the targeted rate of decline! As the study claims, NRHM has not achieved the desired impact on health output either. It is worth noting that rural healthcare is in such a bad shape that the usual practice is for the patients to bring their own bed sheets for treatment in community health centres. Can the urban elite even imagine something like this? The study concludes that though this ambitious mission is first of its kind in the country, much more needs to be done.

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