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BOOK REVIEW

Chromosomes came first

First Years of Human Chromosomes. The Beginnings of Human Cytogenetics

Edited by Peter S Harper. Scion Publishing Ltd, 2006. GBP 33.25 (Hardcover).
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When prominent scientists retire, it is not uncommon that they become interested in the history of their field, especially, when they witnessed decades and heard from their teachers about the period before their active involvement in the field. Peter Harper, a prominent clinical geneticist with special interest in neurology and former chairman of the Medical Genetics Department at the University of Cardiff, Wales, is predestined in many ways for such an activity. Through his position and activities, and also as a former editor of the *Journal of Medical Genetics*, he knows the international human genetic scene very well, and being outgoing and sociable, he does not have major difficulties in obtaining further information from many specialists. The fact that he decided to review the early years of human cytogenetics, a field in which he was never scientifically active, reflects his ongoing scientific curiosity and the obvious need to work in a field, which is now more and more is overshadowed by the rise of molecular genetics, although it laid the basis for the latter.

The author decided to interview the pioneers in cytogenetics, and fragments of those interviews are presented word for word in the book. This approach has a particular strength – it makes the reading easy, exciting and diversified. Harper just came in the last moment as several of the pioneers passed on shortly after the interview (Polani and Therman) and for some of them, he came too late (Lejeune, Hsu, de Grouchy and Schmid). Another aspect worth mentioning is the excellent pictures of prominent cytogeneticists and their work.

Among the less well-known facts, which this book brings to light is the prominent role of Russian cytogeneticists before the regrettable total decline of this field during the Lyssenko era. How some important discoveries were achieved by chance or from a primarily erroneous approach, how psychological factors hampered new developments and how ideology made some excellent researchers blind are some of the fascinating factors (eg, the opinion of Harnden that Australian aboriginals would have a chromosome number different from Caucasians). He also emphasizes the fact that what today gets more and more effaced is the vital importance of cytogenetics for the rapid development of human genetics in general. And he points out the importance of female scientists for the early development of the field, especially when compared to other specialities.

The book covers most important aspects of early cytogenetics. The reviewer misses, for example, some notes about chromosome autoradiography, which was the only way to distinguish some chromosomes within the same group before the advent of banding techniques and laid the basis for the distinction between Cri du Chat and Wolf–Hirschhorn syndromes. A chapter on 'chromosome breakage syndromes' and the early history of the discovery of the 'fragile X syndrome' would also have been warranted. And I would like to appreciate the role of technicians who sometimes performed the important work published by scientists, and however was not even acknowledged in any publication.

Errors? I found some although not of major importance. For example, while the Table on page 156 cites the first two papers on the 4p-syndrome correctly, on the following page, the author cites that Hirschhorn identified the 5p- (and not the 4p-) syndrome. The text also evokes that this author was the first to demonstrate that the distal deletions of 4p and 5p cause two different clinical patterns. In fact, however, this honour is owed to U Wolf who, by applying autoradiography, demonstrated the 4p involvement in his case, while Hirschhorn, even in his second paper, that is, 1965 paper, assumed this only after knowing Wolf's discovery, without yet performing the necessary investigation in his case. On page 158, he mentions 'the 45X Turner karyotype' but it is not correct cytogenetic nomenclature (it should read 45,X), and as the first report on this clinical condition had come from Ullrich years before Turner published her paper, it is nowadays mostly agreed to name this syndrome Ullrich–Turner syndrome.

A local comment from Zurich: Harper mentions that long before Lejeune discovered that Down syndrome is caused by trisomy of chromosome 21, several other researchers, for example, Waardenburg and Davenport, had already assumed that this frequent condition could be due to a chromosome aberration. In fact, the prominent Swiss paediatrician Guido Fanconi had not only proposed the same idea; more specific than the two aforementioned researchers albeit in error, he assumed a monosomy and not a trisomy.[1] Although Fanconi is famous for his reports on cystic fibrosis, Fanconi anemia and others, he himself considered this as the most important idea in his scientific career.[2] It would have been nice and fair to mention this, and it

would not do any harm to the importance of the two other contributors.

In summary, this is a book that appears at the right moment. The author must be honoured for his attempt to prevent many important facts and details in the history of early cytogenetics from being lost forever. The book reads easily; it can be highly recommended for geneticists not only in cytogenetics but also in any field■

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2 Fanconi G: Weitere Fälle von wahrscheinlich eineiigen Zwillingen, von denen der eine gesund ist, der andere einen Mongolismus zeigt. *Helv Paediatr Acta* 1962; **17**: 490–491.

Building Genetic Medicine

Building genetic medicine, a tale of two countries

'Building Genetic Medicine: Breast Cancer, Technology and the Comparative Politics of Health Care' Edited by Shobita Parthasarathy
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This is an interesting book, which describes the development of genetic testing for breast cancer predisposition through a social and public health perspective. These data are comprised of structured interviews as well as information gathered from articles published in newspapers, magazines and scientific journals.

S Parthasarathy provides a historical description of genetic testing for breast cancer in the United States and the United Kingdom. She adopts a neutral tone as she details the divergent approaches in these two countries. The historical and sociological background provided allows the reader to understand how these two countries produced such different systems. Comparing public health systems is a controversial topic and adding the complexity and ethical issues of genetic testing renders this debate more fascinating. The neutrality of the author allows the reader to appreciate the respective strengths and weaknesses of the two systems. For those who are not familiar with the United Kingdom and United States genetic testing strategies, the differences are striking. In the United Kingdom, testing is provided through a structured health service system aimed at reaching public health goals with a particular concern on full coverage of the population. Contrastingly, the United States has a minimally regulated environment in which one biotech company markets genetic testing to consumers and professionals. The author describes how the variety of approaches initially available in each country was eventually replaced by one system. In the United States, Myriad Genetics used its financial strength and intellectual property to drive other providers out of the market. In Britain, proponents of a national BRCA-testing strategy pressured regional genetic clinics to adopt their system. The author describes how, once Myriad established itself as the sole provider in the United States, it failed to expand in Europe where health care professionals, patients and intellectual property rights are viewed in a radically different way. One of the main shortcomings of this book is the absence of data showing what these systems have achieved. The author mentions that as of fall 2006, Myriad had sold 100 000 tests in the United States. How many individuals with breast cancer were identified? One might assume that Myriad tests, which were directly marketed to patients, may have had a lower positive yield since the criteria for identifying women who should benefit from the test were less stringent. These data, however, may not be available.

Overall this book is very thought provoking and demands readers to consider the directions we want our health systems to go. The divergent evolution of BRCA1 and 2 testing in the United Kingdom and the United States should serve as a paradigm to help us prepare for the next revolution: 'whole genome' testing. Biotech companies are racing towards the 1000-dollar genome test, which will raise far more complex medical, ethical and legal issues■

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