The Galton Institute evolved about 10 years ago from the National Institute of Eugenics which was established at University College London from a bequest of Francis Galton (1822–1911). As stated by the cover blur, the "Galton Institute is a learned society founded in 1907. Its four hundred members are drawn from a wide range of disciplines including the biological and social sciences, medicine, law, and administration. One-fifth live abroad. The Institute holds an annual conference and publishes the proceedings. It also publishes a quarterly newsletter. Membership of the Institute is open to all who support its aims" [which are not stated in the blurb].

The current president of the Galton Institute, Professor J S (Steve) Jones of UCL, is definitely not a eugenicist but is an effective communicator and leading ambassador for human genetics to the public.

Victor A McKusick

Myotonic Dystrophy


As with the second edition of this book, published back in 1989, this third edition is an excellent account of clinical and scientific aspects of myotonic dystrophy. The book is easy to read, sustaining this reviewer's attention to scan the whole book in a single sitting, while simultaneously being of value as a reference for closer consultation on specific clinical and scientific details. As stated in the preface, the book is aimed at "clinicians who encounter myotonic dystrophy in its numerous aspects and who need a broad overview of all the different areas" and "research scientists . . . trying to interpret . . . their work in terms of the broader effects on the disease". The book is aimed well. "Broad overview", "broader effects"—myotonic dystrophy involves a variety of clinical systems and presents to a variety of physicians, and the value of this book is in this wide scope, to inform about clinical and scientific areas.

The book is divided into 14 chapters. All but one of these is written by Peter Harper, thus ensuring continuity of style. The Introduction and Background gives an excellent overview of the disorder, noting the "avoidable catastrophes" that are known to occur in myotonic dystrophy. There follows an excellent chapter on the clinical features of the disease, written by one with immense clinical experience. Advice such as examining for myotonia, even when patients deny its existence, and the section on social aspects of disability in the disorder are particularly pertinent. The chapter on other myotonic disorders gives a useful overview of the multiple medical and genetic disorders of proximal myotonic myopathy (PROMM), with a useful table comparing the clinical features of this with those of myotonic dystrophy and with helpful clarification on the nomenclature of DM1 and DM2 in relation to the differing phenotypes. There then follows a series of chapters outlining the clinical features resulting from involvement of specific systems, including smooth muscle, the brain, the eye, and the endocrine system. These chapters are a mine of useful information. Figures such as "25% of patients consider their gastrointestinal symptoms to be the most disabling feature of the disorder" stuck with this reviewer. There is a clear table detailing practical recommendations for surgery and anaesthesia in myotonic dystrophy, both pre- and postoperatively. The sections on the complications of pregnancy in the disorder are similarly clearly presented. A separate useful chapter follows on the disorder in infancy and childhood.

It is not until one is three-quarters of the way through the book that the chapter on the genetic basis of myotonic dystrophy is reached. This is the area in which huge advances in understanding have been made since the last edition of this book. With the clinical chapters, this is a lucid account of the finding of the unstable CTG repeat sequence in the 3′ untranslated region of the myotonic dystrophy protein kinase gene. This chapter and that following on “Molecular and cellular biology of myotonic dystrophy” are concise overviews of the recent molecular biological advances. They cannot be comprehensive but rather give an excellent grounding for those who want to delve into the more detailed scientific publications.

And what of the down sides of the book? Criticisms here relate to the publication style rather than the content. The general quality of the illustrations is fairly poor, with all figures and photographs in black and white. There is oddly shaded in several of the clinical photographs. The reproduction of the CT and MRI images is poor. The histological slides shown would be greatly enhanced if they were in colour. In many places the figures and legends are several pages removed from the appropriate text, making smooth reading awkward. A list of the abbreviations used throughout would be useful. More diagrams in the sections detailing molecular mechanisms would be helpful.

These “publication” criticisms, however, are minor and stand in stark contrast to the excellent text. Yes, a book is always going to suffer the drawback of delay in time to publication. Advances in the molecular understanding of myotonic dystrophy have competed for commercial apace since this volume was published last year, not least with the exciting discovery of CCTG expansions within ZNF9, the DM2 gene, on chromosome 3q21 in August 2001. This does not mean that a volume such as this is obsolete. Far from it. The background, detailed clinical information and, I return to this word again, “broad” overview given in this work, provide an excellent, readable, and retainable account of myotonic dystrophy. I highly recommend this book.

Karen E Morrison

Oxford Medical Databases:
London Dysmorphology


Most clinical geneticists are familiar with the Oxford Medical Databases and new versions...
is possible to configure the display to the style of the previous version with buttons instead of the toolbar. Other novel features of this version include major changes in the photo card and the photographer viewer. These include the ability to change the size of the thumbnails in the imagelinks in the Photo Library, display several or all images for a syndrome simultaneously for comparison, and import selected images of a syndrome from the Photo Library to a personal collection of images called “My Collection”. It is also possible for users to import clinical photographs of their own patients into “My Collection”, thus allowing comparisons to be made with the images in the Photo Library. Most picture file formats are supported, such as JPEG, Windows Bitmap, and Tagged Image File Format, making this a very useful function. There are also links from the Databases to Online Mendelian Inheritance in Man (OMIM) and Medline in case the user wishes to obtain more information about an individual syndrome or reference.

Another important feature of version 3.0 is the ability to download regular updates to both the LDDDB and LNDB online, at no extra cost. This will allow the Databases to remain up to date with all recent advances, especially with respect to identification of genes for well recognised syndromes in the creation of new dysmorphic and neurogenetic syndromes.

Both Databases are relatively easy to use, even for first time users. However, a working knowledge of clinical neurology and neurology is necessary to harness their full potential and in order for these Databases to be used effectively. It is difficult to find fault with any aspect of the Databases. The only drawback is the price of the combined package (£1595 + VAT for the single user version and £1125 + VAT for the single user upgrade). Although the Databases and Photo Library can all be purchased individually, the cost of each component (£595 + VAT for the single user version and £425 + VAT for the single user upgrade) is still prohibitive and will dissuade most clinicians from buying a copy for their personal use.

The clinical genetics community owes a huge debt of gratitude to the two authors of these Databases. It is difficult to envisage the practice of clinical dysmorphology and neurogenetics without access to the LDDDB, LNDDB, and Dysmorphology Photo Library. For most clinical geneticists faced with an unknown dysmorphic or neurogenetic syndrome, these Databases are likely to be the first port of call. A thorough search of the Databases will often suggest a diagnosis for such patients. It is this reviewer’s firm belief that no clinical genetics or neurology department can afford to be without these Databases. Perinatal pathologists and the public will also benefit enormously from access to them.

Mohnish Suri

Fragile X Syndrome - Diagnosis, Treatment and Research


This is the third edition of a book that is already well known to clinical geneticists and genetic counsellors. Probably, just as many scientists from the cytogenetic and DNA laboratories are familiar with the title. The first edition was printed in 1991, at the same time that the mutation that causes fragile X syndrome was identified. When the second edition appeared in 1996, more information about the gene product, FMRP, Advances in understanding molecular and cellular changes in fragile X syndrome have proceeded, but in smaller increments in more specialised fields. The neuroscientific aspects of the syndrome are thus allotted more space in this new edition and, perhaps related to this, there has been an editor substitution with Randi Hagerman, spouse of Paul and molecular arm of the husband-wife collaboration, replacing Amy Cronister, who continues to make a major contribution co-authoring the chapter on genetic counselling.

To make room for new information on neurosciences, the chapter on cytogenetics of fragile X syndrome has been cut but, really, this omission should be regarded as a sound reason for retaining the second edition on the bookshelf. Happily, the opening chapter by Randi Hagerman is not shortened as it contains a wealth of clinical information including perfect illustrations of macroorchidism and the value of orchidometer. Older geneticists will wistfully recall epic estimations of testicular volume in burly men pre-1991, but new trainees need only gaze in astonishment at these pictures and perhaps give silent thanks for the trimneocolicotide repeat.

The essays in this edition also provide a good illustration of how one dramatic discovery, the CGG expansions, was previously perplexing, but also leads to many more complex questions being posed. Ted Brown's very clear chapter on the molecular biology of the fragile X mutation sits alongside Stephanie Sherman's fascinating account of the syndrome's epidemiology, but self-congratulatory feelings about one's adult learning capacity are then dashed by detailed state of play reviews of protein studies, an animal model, and the brain structural phenotype. After this, a review of the neuropsychology of the syndrome brought home to me the importance of a technical approach to the definition of behavioral stereotypes, and wound up the first half of the book.

In the second half, essays explore treatment options and give practical advice on dealing with learning difficulties and troublesome behaviours. Much of the best advice is of a general nature and is not specific to the management of children with fragile X syndrome. The chapter on drug therapies will also be of interest to the paediatrician or specialist in learning disability who supervises clinical management. The accounts of academic and psychological interventions that may improve self-concept and self-esteem of people with fragile X should be beneficial to the families and so much information that is of value to teachers and therapists. To close, there are useful appendices with web and e-mail addresses for general information, educational software, and other helpful resources.

In summary, this is a book that may be read by the fireside; especially pleasing are the many paragraphs that supply historical back- ground to major discoveries about the syn- drome, but the book's main use is for reference purposes. Quite simply, it should always be consulted when considering clinical problems.

John Tolmie