Belated appreciations

Martin Bobrow

The History of a Genetic Disease: Duchenne Muscular Dystrophy or Meryon's Disease. By Alan E. H. Emery and Marcia L. H. Emery. *Royal Society of Medicine, 1, Wimpole Street, London WIM 8AE, UK: 1995. Pp. 248. £20.*

DUCHENNE muscular dystrophy is in many ways the perfect model of human monogenic disease. This enjoyable short book brings out many interesting facets. The disorder has a highly characteristic phenotype, but was not fully recognized as a separate clinical entity until the second half of the nineteenth century. It has a classical X-linked recessive pattern of inheritance, only males being affected (with very rare exceptions), whereas unaffected women transmit the disorder. This too was not appreciated until the last quarter of the nineteenth century, more than 25 years after the clinical and pathological features of the disorder were clearly described, which is particularly puzzling because the same familial pattern of haemophilia was known from Talmudic times and formally described in 1803. All these and many other entertaining facts are covered — although with little speculation on explanations for these puzzles — in the early chapters of the Emerys' book.

The driving motive for the authors seems to have been a desire to see more credit given to Edward Meryon, who published clear clinical descriptions of the disorder, with remarkably prescient pathological observations: "... the sarcolemma or tunic of the elementary fibre was broken down and destroyed". That was in 1852, nearly ten years before the early publications of Guillaume Benjamin Amand Duchenne, the French neurologist whose name has been associated with the disease ever since. Why was Meryon's work buried? The hypothesis here is that Meryon was a retiring person, who published his important observations in one paper, with a short book chapter 15 years later. The energetic Duchenne published frequently, lectured often and generally made sure people knew of his work. Again, the disease provides illustrations more than a century old of issues still current today. That the Emerys think a lot of Meryon is evident from the table of contents. From 205 pages of text, he gets 30 pages to Duchenne's 19, and 24 for the whole of modern molecular genetics.

This fascination with Meryon, Duchenne and their predecessors and contemporaries infuses the first half of the book, making delightful reading. Discursive background material, to set the issues in proper social and historical context, adds to the pleasure. Where else could I have learned that Duchenne designed a version of the modern muscle biopsy needle, which he kept from rusting by storing it in alcohol, thus accidentally avoiding problems of sepsis, about the causation of which he could have known nothing? The distinguished anatomist and surgeon Sir Charles Bell owned a building that subsequently became the Lyric Theatre in London's West End. It was a school of anatomy then, "the present stage door formerly being the entrance for 'subjects for dissection'".

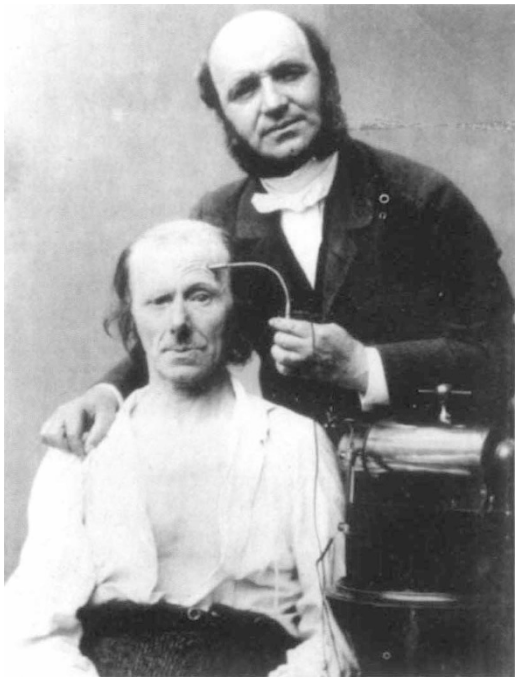
From *The History of a Genetic Disorder*

Meryon: astute but retiring observer.

The authors give appropriate and welcome credit to Newton Morton, who introduced discriminant and segregation analyses into modern human genetics as part of a large population study of Duchenne muscular dystrophy. Tony Murphy used the disease as the basis for developing Bayesian risk-analysis procedures. These contributions have long since permeated much of clinical genetics.

Muscular dystrophy also provided the basics for a very different development. The Muscular Dystrophy Association was formed in the United States in 1950, and the Muscular Dystrophy Group in the United Kingdom in 1959. They were forerunners of the enormously important movement for patient and family advocacy groups, to raise funds for care and research and to maintain political pressure on behalf of those suffering from relatively uncommon diseases.

The latter half of the book lacks, to my mind, the same drive and focus. Various other muscular dystrophies are described at a level of clinical detail that strays perilously close to a basic medical text rather than an historical account. The massive fundamental advances of the past decade are superficially described, liberally laced with thumbnail sketches of a selection of the important contributors. These are constructed with sufficient similarity to make reading them somewhat like leafing through a pile of applicants for an appointments committee — although it would be a very lucky committee to have drawn quite this group of applicants.



From *The History of a Genetic Disorder*

Duchenne: energetic self-publicist.

On the opening page, the authors write that "... the history of the disease is unique because it encapsulates so many developments in medical science over the last 150 years". I rather suspect that Duchenne muscular dystrophy is not all that unusual, and that the real interest of the story is as an exemplary model for many other biomedical advances. As such, and because of the strong overt commitment to Edward Meryon, at least the first half of the book should appeal to a wider audience than just those interested in muscular dystrophy. □

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