Duchenne Muscular Dystrophy (Oxford Monographs on Medical Genetics)

Alan E. H. Emery, Francesco Muntoni, Rosaline C. M. Quinlivan
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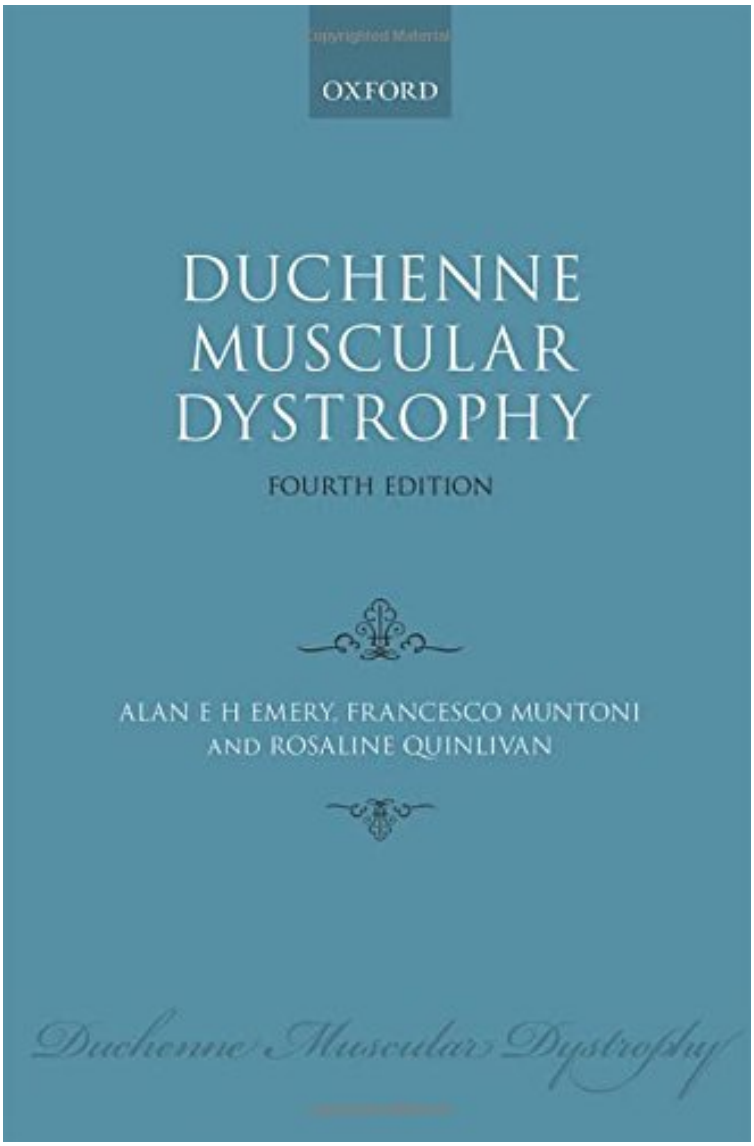

#2554361 in Books 2015-04-26Original language:EnglishPDF # 1 6.40 x .90 x 9.30l, #File Name: 0199681481320 pages | File size: 65.Mb

Alan E. H. Emery, Francesco Muntoni, Rosaline C. M. Quinlivan : Duchenne Muscular Dystrophy (Oxford Monographs on Medical Genetics) before purchasing it in order to gage whether or not it would be worth my time, and all praised Duchenne Muscular Dystrophy (Oxford Monographs on Medical Genetics):

0 of 0 people found the following review helpful. Five StarsBy Lisa J. ZimmermanFantastic0 of 0 people found the following review helpful. Great bookBy Kelsey P.This book was very informative with lots of information regarding

Duchenne. When my 20 month old cousin got diagnosed with Duchenne all I wanted to do was learn more about it.

Duchenne Muscular Dystrophy, an inherited and progressive muscle wasting disease, is one of the most common single gene disorders found in the developed world. In this fourth edition of the classic monograph on the topic, Alan Emery and Francesco Muntoni are joined by Rosaline Quinlivan, Consultant in Neuromuscular Disorders, to provide a thorough update on all aspects of the disorder.Recent understanding of the nature of the genetic defect responsible for Duchenne Muscular Dystrophy and isolation of the protein dystrophin has led to the development of new theories for the disease's pathogenesis. This new edition incorporates these advances from the field of molecular biology, and describes the resultant opportunities for screening, prenatal diagnosis, genetic counselling and from recent pioneering work with anti-sense oligonucleotides, the possibility of effective RNA therapy. Although there is still no cure for the disorder, there have been significant developments concerning the gene basis, publication of standards of care guidelines, and improvements in management leading to significantly longer survival, particularly with cardio-pulmonary care. The authors also investigate other forms of pharmacological, cellular and gene therapies.Duchenne Muscular Dystrophy will be essential reading not only for scientists and clinicians, but will also appeal to therapists and other professionals involved in the care of patients with muscular dystrophy.

"A highly specialized book like this requires expertise and a unique perspective, and there are no comparable books. The authors have been able to present the information concisely and simply, resulting in an easy to read book. It would take seasoned medical geneticists and neurologists little time to review it. Weighted Numerical Score: 98 - 5 Stars!" -- Luis F. Escobar, Doody'sAbout the AuthorAlan E. H. Emery, Emeritus Professor of Human Genetics, University of Edinburgh, Honorary Fellow, Green Templeton College, Oxford and Honorary Visiting Fellow, Peninsula College of Medicine, Plymouth, UK,Francesco Muntoni, Professor and Honorary Consultant in Paediatric Neurology, UCL Institute of Child Health and Great Ormond Street Hospital Foundation Trust, London, UK,Rosaline C. M. Quinlivan, Consultant in Neuromuscular Disorders, Centre for Neuromuscular Diseases, National Hospital for Neurology and Neurosurgery, and Dubowitz Neuromuscular Centre, Great Ormond Street Foundation Trust, London UKAlan EH Emery is a qualified physician, scientist and educator with wide experience of patient care and human genetics laboratory research. In 1966 he was first to describe a unique form of Emery-Dreifuss muscular dystrophy and to discover a significant biochemical defect linked to the pathogenesis of Duchenne muscular dystrophy. On this he wrote the first detailed scientific monograph in 1987 (Duchenne Muscular Dystrophy, Oxford University Press; 4th edition, 2014) and in 1989 founded the European Neuromuscular Centre to research related disorders. He has published over 400 scientific papers and written or edited 30 books regarding clinical, biochemical and genetic studies in neuromuscular disorders. For his work over the last 40 years he has received many national and international awards, including the Lifetime Achievement Award of the World Federation of Neurology. He is currently a Vice-President of the Muscular Dystrophy Campaign of Great Britain.Francesco Muntoni specialised in Child Neurology and Psychiatry in Italy before moving to England in 1993. From 1993 he worked at Hammersmith Hospital's Neuromuscular Centre under the direction of Professor Victor Dubowitz and after 1996 as the Centre's Research and Clinical Director. In 2007 he moved with the clinic, pathological and research team to Dubowitz Neuromuscular Centre, UCL Institute of Child Health Great Ormond Street Hospital (GOSH). He continues his clinical and research activities as Lead for the Nationally Commissioned Service on Congenital Muscular Dystrophies and Congenital Myopathies, Neuroscience Theme Lead for Institute of Child Health, and the Novel Therapies Lead for the GOSH Biomedical Research Centre. He is also co-director of the MRC Translational Research Neuromuscular Centre at UCL. In 2011 he received the Premio 'Navicella' for Medical Science Achievements at Castelsardo, Italy, and in 2013 became a Vice-President of the Muscular Dystrophy Campaign of Great Britain.Ros Quinlivan trained at University College London, initially in Psychology and, subsequently, medicine. Her post-graduate training was within London Teaching Hospitals. She has been a Consultant in Neuromuscular Disease for 18 years and has wide clinical experience of both paediatric and adult onset genetic muscular disorders. She is currently the Clinical Lead for Transition for Young Adults with Neuromuscular Disease at Great Ormond Street Hospital and The National Hospital for Neurology and Neurosurgery. She leads the Nationally Commissioned Service for McArdle Disease and related disorders and is joint co-ordinating editor for the Cochrane Neuromuscular Disease Group. In 2010 she was named the Muscular Dystrophy Campaign's 'Clinician of the Year', and is currently an editorial board member for the journal 'Neuromuscular Disorders'.