Oxford Desk Reference: Clinical Genetics and Genomics (Oxford Desk Reference Series)

Helen V. Firth, Jane A. Hurst
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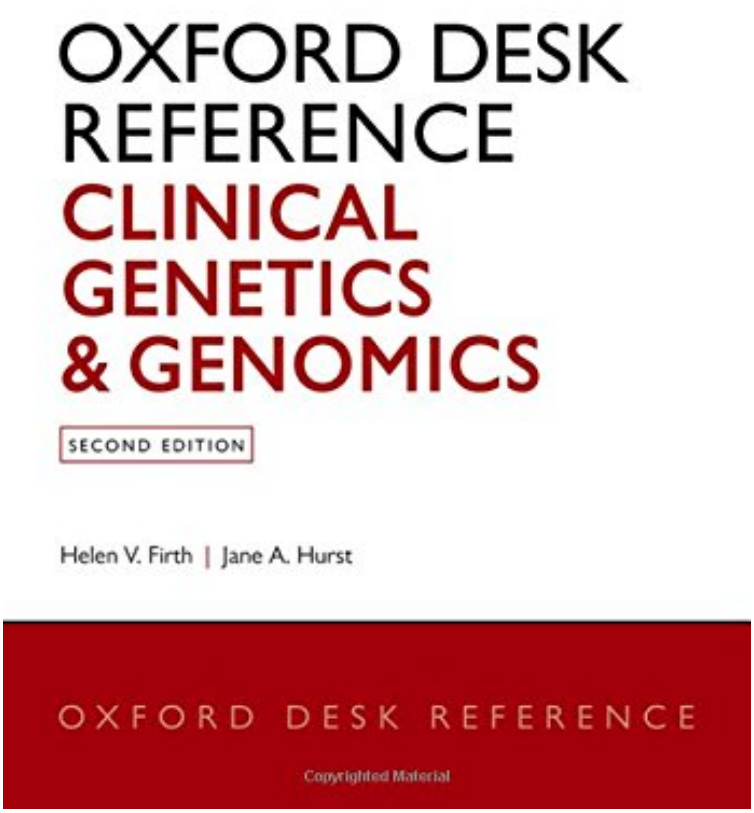

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Helen V. Firth, Jane A. Hurst : Oxford Desk Reference: Clinical Genetics and Genomics (Oxford Desk Reference Series) before purchasing it in order to gage whether or not it would be worth my time, and all praised Oxford Desk Reference: Clinical Genetics and Genomics (Oxford Desk Reference Series):

A popular and easy-to-use guide, this book is a must-have tool for clinical consultations in genetics and genomic

medicine.Ideal for quick reference during practice, it covers the process of diagnosis, investigation, management, and counselling for patients. With a strong evidence base and international guidelines, it puts reliable and trustworthy guidance at your fingertips. Designed for use as a first-line guide, the A to Z format ensures it's accessible, and the simple layout makes it easy to assimilate information. Highly illustrated, the book also contains up-to-date glossaries of terms used in genetics and dysmorphology providing quick reference for key concepts. The second edition is an eagerly anticipated update of the gold standard in the specialty. It covers new developments in the field, particularly the advent of genome-wide sequencing and major updates in cancer. Fifteen new topics have been added, including Sudden cardiac death, Neonatal screening, and Ciliopathies. The authors have used their experience to devise a practical clinical approach to many common genetic referrals, both outpatient and ward based. The most common Mendelian disorders, chromosomal disorders, congenital anomalies and syndromes are all covered, and where available diagnostic criteria are included. In addition there are chapters on familial cancer and pregnancy-related topics such as fetal anomalies, teratogens, prenatal and pre-implantation diagnosis and non-invasive prenatal testing. The book also provides information on the less common situations where management is particularly complex. Both practical and pertinent, Oxford Desk Reference: Clinical Genetics and Genomics is the companion you need by your side during clinical consultations.

from previous edition: "It is very refreshing to review a book written for clinicians by clinicians, which is in a format that reflects situations actually encountered in practice. Information provided by the referring doctor to a clinical geneticist or other specialist before a clinicor ward consultation is usually limited. This new text takes common referral indications and, in a standardized format that manages to be brief and clear without skimping on detail, reminds the clinician of diagnostic possibilities and strategies for investigation and management. This will allow thebest possible use to be made of an individual consultation by both the patient and the doctor." --Dian Donnai, Professor of Medical Genetics, University of Manchester, Consultant Clinical Geneticist, Regional Genetics Service, St Mary's Hospital, Manchester, UK"I have been impressed with the thoughtfulness of the topics. This should be a great help to many people who are part of the clinical genetics team....There are up-to-date summaries for the staff member who needs a refresher, as well as the glossary and the headings on fundamental topics, likeAD inheritance, for those just starting out." --Lewis B. Holmes Professor of Pediatrics, Harvard Medical School and Chief, Genetics and Teratology Unit, Massachusetts General Hospital for Children, Boston, Massachusetts, USA"This is going to be an extremely useful reference source. The authors have done an outstanding job of summarizing, in one or two pages, pertinent recommendations regarding diagnoses and management of specific disorders as well as practical approaches to a variety of problems that commonlypresent in real life." --Marilyn Jones, Adjunct Professor of Pediatrics, University of California, San Diego and Director, Dysmorphology and Genetics, Children's Hospital San Diego, USA"...a comprehensive and highly focussed guide to clinical genetics that should certainly rank as an indeispensable handbook for consultants in clinical genetics, genetic counsellors and paediatricians. However, it should also be extremely useful for PhD students in nearly all disciplineswithin medical and/or human genetics. Its major strength is the well-conceived and clearly laid out format which enables the reader to obtain a rapid yet quite substantial overview of a plethora of difficult topics..." --Human Genetics Oct 07"The authors of [this book] deserve to be congratulated for achieving the impossible...Overall this book is a winner and is a must for every clinical genetics department. This is arguably the most important book ever published for trainees in genetics...[but] can be considered as an extremelyuseful reference source to any genetics physician...this book is a 'peripheral brain' and 'lifesaver' for geneticists in many situations!" --Ulster Medical Journal Vol 75, no 3 Dec 2006"If there was a Booker Prize for new texts on clinical genetics, then the winner this year would be a foregone conclusion. No one else could possibly come up with and entry as good as this. ... the definitive hands-on guide to clinical genetics. ... The breadth and depth of informationprovided is remarkable. ... As a practical guide to the specialty of clinical genetics this book has no match, and overall it represents an awesome achievement. How did the authors manage to acquire and collate all this knowledge? Where did they find all this information? ... If your department canonly afford one book this year, make it this one. Better still, buy your own copy and keep it hidden because it is going to be much in demand." --BMJ May 2006"This is an amazing compilation of genetic knowledge. It provides a fantastic tool for clinical geneticists who require a fast review of specific genetic subjects while performing clinical consultations. ... Condensation of the amount of information included in this wonderful book could not bedone any better. ... This is a most-have tool for all clinical geneticists who require quick and specific reviews in clinical practice. ... Dr Firth and Hurst have achieved a tremendous goal. They have been able to summarize a tremendous amount of information in clinical genetics and convert it toan excellent tool for the practice of the specialty. It could not be done any better. The magnificent work done suggests that as the field of clinical genetics expands, further editions will be needed. This is a must have book, and a second edition would be expected." --Doody's Journal March 2006About the AuthorHelen V. Firth, Consultant in Clinical Genetics, Cambridge University Hospitals, Cambridge, UK and Hon Faculty Member, Wellcome Trust Sanger Institute, Hinxton, UK,Jane A. Hurst, Consultant in Clinical Genetics, Great Ormond Street Hospital, London, UKDr Helen Firth, DM FRCP DCH is a Consultant Clinical Geneticist at Cambridge University Hospitals, an

Honorary Faculty Member of the Wellcome Trust Sanger Institute, and a Bye-Fellow of Newnham College, Cambridge. Her main research interests are in mapping the clinical genome and the matching of rare genomic variants to empower discovery and diagnosis in rare disease. In 2004, she initiated the DECIPHER project (http://decipher.sanger.ac.uk) that enables clinicians and scientists around the world to share information about rare genomic variants to facilitate diagnosis and help to elucidate the role of genes whose function is not yet known. In 2010 Dr Firth became Clinical Lead for the Deciphering Developmental Disorders study (DDD study) (http://www.ddduk.org), one of the world's largest nationwide, genome-wide sequencing projects in rare disease. The study aims to improve diagnosis and further understanding of the genomic architecture of severe developmental disorders.Dr Jane Hurst is a clinician working full time as a clinical geneticist in the one of the leading children's hospitals in the world; a centre of excellence for the diagnosis and treatment of rare diseases. She moved to her current post in 2010 to lead the dysmorphology service after 18 years working in Oxford, UK.Although primarily a patient-focussed clinician, she has always worked closely with scientific colleagues by identifying families that give important clues to the genetic aetiology. Thus early in her career she identified the first family shown to have leptin deficiency and the two families that led to the cloning of the FOXP2 gene.