

A review of ocular genetics and inherited eye diseases

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Abstract

During the past twenty years, there has been an exponential increase in the knowledge and understanding of ocular genetic diseases and syndromes. The number of human eye diseases that have a known genetic or hereditary component continues to increase. In addition, genetic diseases are the most common cause of blindness in infants and children in developed countries. Optometrists are likely to encounter patients with inherited eye disorders. They may be the first clinician the patient consults. Inherited eye diseases may be isolated (only affecting the eye) or part of a complicated syndrome. Both isolated eye diseases and genetic syndromes can have identifiable gene mutation known to cause the disease.

Knowledge of the clinical and molecular features of ocular genetics and inherited eye diseases is important for appropriate diagnosis and patient management. This article reviews the current information on ocular genetics and inherited eye diseases. The ocular conditions described in the review have significant visual impairment and blindness consequences. Therefore, optometrists (as the mostly likely first line of consultation) should be able to diagnose the condition appropriately first before they could make any management, care or referral plan. Visual aids are, of course, one of the management options for such patients with visual impairment. (*S Afr Optom* 2012 71(4) 178-189)

Key words: Ocular genetics, gene, genome, mutation, Mendelian genetics.

Introduction

The interest in ocular genetics by ophthalmologists who described the patterns of inheritance of familiar eye diseases contributed much to the human discovery of ocular genetics. The pace of *genes* (units of heredity) discovery has increased a lot and we now have the Human Genome Database¹⁻⁴. Of the approximately 4000 genetic diseases and syndromes known to affect humans, at least one-third involve the eye³⁻⁶. Over the last two decades, there has been an exponential increase in our knowledge of genetic eye diseases and syndromes. The purpose of this paper is

to provide a review of the current understanding of ocular genetics. This may be useful to clinicians and researchers in their ability to provide accurate genetic diagnosis and appropriate counselling to patients and their families.

Basics of genetics

Genetics is the study of hereditary and the variation of inherited characteristics⁶. It is a matter of general observation that the cubs of a lion resemble the lion and pups of a dog resemble the dog not only in shape but also in habits, strength and func-

Genetics In Ophthalmology A Catalog Of Inherited Eye Diseases

Saul Merin



Genetics In Ophthalmology A Catalog Of Inherited Eye Diseases:

National Library of Medicine Current Catalog National Library of Medicine (U.S.), Molecular Genetics of Inherited Eye Disorders Alan F. Wright, 1994-12-01 Molecular Genetics of Inherited Eye Disorders provides an authoritative and up to date account of molecular genetic advances in a wide spectrum of genetic eye disorders and forms the second volume in the Modern Genetics book series The field has produced some dramatic and often unexpected findings in recent years ranging from the elegant unravelling of the molecular basis of colour vision defects to the subtle complexity of the retinoblastoma gene The role of crystallins in congenital cataract and of the rhodopsin molecule in retinitis pigmentosa are discussed illustrating the importance of the candidate gene approach to genetic eye disease Reverse genetic approaches to the cloning of genes responsible for aniridia and choroideremia exemplify the power of the new genetic techniques and signal the start of the next experimental phase in which the functional characterization of identified genes begins Current Catalog National Library of Medicine (U.S.), 1983 First multi year cumulation covers six years 1965 70 **Genetic Diseases of the Eye** Elias I. Traboulsi, Professor of Ophthalmology and Director Center for Genetic Eye Diseases Elias I Traboulsi, Med, Virginia Miraldi Utz, Department Chair Pediatric & Neuro-Ophthalmology Arif O Khan, 2025-08-22 Since the publication of the Human Genome Project in 2001 and the development of Genome Wide Association Studies GWAS there has been a major shift toward large scale international collaboration in genetic eye disease research Eye researchers involved in genetics have now joined forces with traditional epidemiologists to develop the field of genetic epidemiology Whilst ophthalmic geneticists had focused on rare pathogenic variants mutations with strong effects epidemiologists had focused on common gene variants with small but additive effects see Figure 1 These two areas have now merged identifying a spectrum of genetic variants that are rare intermediate or common with strong medium or mild pathogenicity *Ophthalmology, E-Book* Myron Yanoff, Jay S. Duker, 2022-11-06 Through five highly regarded editions Ophthalmology by Drs Myron Yanoff and Jay S Duker has remained one of the premier texts in the field providing authoritative guidance on virtually any ophthalmic condition and procedure you may encounter The fully revised 6th edition of this award winning title continues to offer detailed superbly illustrated content from cover to cover with extensive updates throughout to keep you current with the latest advancements and fundamentals throughout every subspecialty area in the field An easy to follow templated format convenient single volume and coverage of both common and rare disorders make this title a must have resource no matter what your level of experience Offers truly comprehensive coverage including basic foundations through diagnosis and treatment advances across all subspecialties genetics optics refractive surgery lens and cataract cornea retina uveitis tumors glaucoma neuro ophthalmology pediatric and adult strabismus and oculoplastics Features streamlined templated chapters a user friendly visual layout and key features boxes for quick access to clinically relevant information and rapid understanding of any topic Contains four new chapters covering Phototherapeutic Keratectomy IOL Optics Bag in the lens Cataract Surgery

and Capsulectomy Modern devices apart from FLACS Includes a fully revised and updated chapter on refractive surgery screening and corneal imaging as well as an expanded chapter on corneal cross linking Provides up to date information on the latest advances in the field including new therapies for retinoblastoma such as intravenous and intraarterial chemotherapy less common retinal tumor simulators of retinoblastoma OCT Angiography glaucoma stents new drug delivery platforms IOL optics phototherapeutic keratectomy intraocular pressure monitoring and more Includes more than 2 000 high quality illustrations and an expanded video library with more than 60 clips of diagnostic and surgical techniques including new videos of nystagmus Contains updated management guidelines for central retinal artery occlusions CRAO Provides fresh perspectives from new section editors Drs Carol Shields and Sumit Sam Garg Enhanced eBook version included with purchase Your enhanced eBook allows you to access all of the text figures and references from the book on a variety of devices

Genetics in Ophthalmology B. Wissinger, S. Kohl, U. Langenbeck, F. Bandello, 2003-07 The objective of this publication is to enhance mutual understanding and communication between ophthalmologists molecular geneticists genetic counselors and biomedical researchers In the introductory chapter current genetic paradigms and experimental genetic approaches relevant to the nature of hereditary disorders are discussed The following contribution on the epidemiology of hereditary ocular disorders provides an excellent reference to geneticists as well as clinicians Myopia is presented as an example of a complex clinical phenotype where genes and environment interact Further molecular ophthalmogenetic topics such as corneal dystrophies cataract glaucoma optic neuropathy non syndromic and syndromic pigmentary retinopathies defects of vitamin A metabolism and macular dystrophies including age related macular degeneration are investigated in depth The volume concludes with a survey of color vision deficiencies a discussion of animal models and gene therapy and a useful description of technical devices supporting patients who are losing sight

Clinical Ophthalmic Genetics and Genomics Graeme C.M. Black, Jane L. Ashworth, Panagiotis I. Sergouniotis, 2022-01-20 Practical Genomics for Clinical Ophthalmology provides in depth coverage of the clinical applications of genomics in eye disease with a key emphasis on case study based instruction in patient care and genetic counseling aspects genetic and genomic diagnostics and treatment pathways The book presents the latest information on genetic and genomic test results best practices for delivery of results to patients and families and ongoing research into therapeutics with specific chapters covering non syndromic inherited retinal disease syndromic IRD vitreoretinopathies lens abnormalities corneal disease albinism anterior segment dysgenesis glaucoma developmental eye abnormalities nystagmus ocular motility disorders optic neuropathies phacomatoses and retinoblastoma and more In addition clinical case studies illustrate examples of common genetic eye disorders and highlight vital learning points for the reader Presents the work of leading international researchers and clinicians who speak in depth on the clinical applications of genomics in diagnosis and treatment of eye disease Provides full color richly illustrated chapters that cover current genetic and genomic testing methods employed in ophthalmology Includes instructions on the

diagnosis and treatment of a wide range of conditions including non syndromic inherited retinal disease syndromic IRD vitreoretinopathies lens abnormalities corneal disease albinism anterior segment dysgenesis glaucoma and more Contains case studies that illustrate common genetic eye disorders and highlight vital learning points for the reader *Taylor and Hoyt's Pediatric Ophthalmology and Strabismus, E-Book* Christopher J. Lyons, Scott R. Lambert, 2022-05-17 Providing foundational knowledge and expert guidance on the complete spectrum of childhood eye disorders and strabismus Taylor and Hoyt's Pediatric Ophthalmology and Strabismus 6th Edition remains the 1 reference of choice for practicing and trainee pediatric ophthalmologists Edited by global leaders in the field Drs Christopher J Lyons and Scott R Lambert this newly revised volume offers authoritative coverage of all the pediatric ophthalmic conditions you re likely to encounter in practice including the latest clinical advances in etiology diagnosis and medical and surgical management Comprehensive updates as well as new chapters images and video clips make this well regarded title the most current and complete reference available in this evolving field Offers state of the art coverage of key areas such as OCT and current imaging techniques for the eye orbit and visual pathways anti VEGF treatment for retinopathy of prematurity pediatric cataracts childhood glaucoma and minimally invasive strabismus surgery Contains five new chapters covering Imaging the Child s Eye Aniridia Syndromes Mitochondrial Disorders Neurofibromatosis Type 1 and 2 and Myasthenia Gravis Includes a unique practical problems section designed to help you handle difficult patient situations and a how to perform strabismus surgery chapter with extensive step by step illustrations for complete visual guidance Features more than 1 800 high quality images and illustrations including 600 new to this edition that provide visual guidance in diagnosis and management Provides access to more than 50 narrated instructional video clips nine are new depicting multiple diagnostic and surgical techniques including the insertion of prosthetic eyes lensectomy pediatric cataract extraction complications during strabismus surgery goniotomy the removal of conjunctival tumors and more Enhanced eBook version included with purchase Your enhanced eBook allows you to access all of the text figures and references from the book on a variety of devices [Fetal Gene Therapy for Fetal Phenylketonuria](#) Edenilson Brandl, 2025-06-28 It is with great enthusiasm that I present this comprehensive work on the cutting edge of medical science Fetal Gene Therapy for Fetal Phenylketonuria At the heart of this work we explore the fascinating horizons of genetic intervention in fetal development particularly aimed at correcting the manifestations of fetal phenylketonuria Throughout these pages we navigate the intricate intricacies of messenger RNA mRNA injection viral vectors and gene editing technologies such as TALENs and CRISPR Cas9 revealing innovative strategies for expressing or correcting specific proteins in the fetal environment The journey includes not only technological advances but also explores the intersections between advanced fetal medicine legislation ethics and the psychosocial implications of fetal gene therapy However we cannot ignore the significant challenges and complexities associated with genetic manipulation at such a crucial stage of human development Each chapter is meticulously crafted to provide a comprehensive understanding from an

introduction to the fundamentals of fetal gene therapy to future perspectives and potential long term implications This book is intended for researchers healthcare professionals students and anyone interested in delving into the frontiers of modern medicine By sharing these findings and reflections I hope to contribute to the advancement of knowledge and stimulate dialogue about the promising future and ethical challenges associated with fetal gene therapy May this work inspire those who seek to understand and shape the future of fetal medicine Clinical Neuro-Ophthalmology Mr. Rohit

Manglik,2024-03-04 EduGorilla Publication is a trusted name in the education sector committed to empowering learners with high quality study materials and resources Specializing in competitive exams and academic support EduGorilla provides comprehensive and well structured content tailored to meet the needs of students across various streams and levels

Inherited Eye Diseases Saul Merin,2005-08-12 Keeping pace with the changing face of genetics in ophthalmology this Second Edition contains new chapters on molecular genetics thrombophilia and genetically triggered retinal vascular diseases as well as offers many new subchapters highlighting current research by recognized leaders in the field *Genetic Mutations Associated with Ocular Diseases* Minzhong Yu,Rachida Bouhenni,Wei He,Shree K. Kurup,2022-02-04

Ophthalmic Disease in Veterinary Medicine Charles L. Martin,J. Phillip Pickett,Bernhard M. Spiess,2018-09-03 5 Doody s Review 98 100 Featuring a new author team this seminal book has been completely updated and significantly expanded Following an outline of the basic elements of ophthalmic science relevant to study and practice the authors provide a detailed description and discussion of each condition including etiology clinical signs diagnosis prognosis and therapy as well as a section dedicated to presumed inherited eye disorders The focus throughout remains on the clinical needs of the practitioner and the student Beautifully illustrated in full color the book is aimed at trainee and practicing veterinarians interested in all types of ophthalmic diseases and disorders Although the focus is on small animals there is additional material on the horse and the cow For this edition a diverse group of authors from many geographic regions of training and practice offer a breadth of clinical expertise Atlas of Inherited Retinal Diseases Stephen H. Tsang,Tarun

Sharma,2018-12-21 This Atlas of Inherited Retinal Disorders provides a thorough overview of various inherited retinal dystrophies with emphasis on phenotype characteristics and how they relate to the most frequently encountered genes It also meets the previously unmet needs of PhD students who will benefit from seeing the phenotypes of genes they work on and study Further because genetic testing costs are quite high and spiraling higher this Atlas will help geneticists familiarize themselves with the candidate gene approach to test patients genomes enabling more cost efficient testing This invaluable atlas is organized into eight sections starting with an introduction to the basic knowledge on retinal imaging followed by diseases listed according to inheritance pattern and disorders with extraocular manifestations grouped by defining features This structure will be intuitive to clinicians and students studying inherited retinal disorders Pediatric Ophthalmology and Strabismus,Expert Consult - Online and Print,4 Creig S. Hoyt,David Taylor,2012-01-01 Pediatric Ophthalmology and

Strabismus is your one stop source for comprehensive coverage of all the pediatric ophthalmic conditions you are likely to encounter in practice Extensively updated with expert contributions from leaders in the field and now featuring online instructional videos this ophthalmology reference delivers all the state of the art guidance you need to effectively diagnose and manage even the most challenging eye diseases and disorders seen in children Take a holistic approach to patient management that considers the family and ensures optimal doctor patient relationships Get a balanced view of etiology diagnosis and management and access unique guidance on the practical problems encountered in real life clinical cases Impresses the importance of systemic disease in diagnosis and management Apply all the latest clinical advances through updated coverage of strabismus diagnosis management and complications retinal dystrophies imaging AIDS in children developmental biology cerebral visual impairment child abuse severe developmental glaucoma and corneal dystrophies Get rich visual guidance in diagnosis and management from over 1 700 full color illustrations Access advice from the experts with contributions from several new top researchers and clinicians Find the answers you need quickly and easily through a consistent chapter organization and highly accessible clinical information Browse the complete contents of Pediatric Ophthalmology and Strabismus online download all the images and watch brand new procedural videos at www.expertconsult.com

Ophthalmology Myron Yanoff, Jay S. Duker, 2009-01-01 Based on feedback the authors have streamlined their bestselling reference to zero in on just the clinical answers ophthalmologists need in day to day practice This new edition presents unparalleled guidance on nearly every ophthalmic condition and procedure

Certified Ophthalmic Technician Exam Review Manual Janice K. Ledford, 2024-06-01 As the field of eye care has advanced so have the knowledge and skills needed to best care for our patients Certification is a way to hold the profession to a high standard that is appreciated and in some cases required by clinics and offices everywhere For the past 30 years Janice K Ledford s exam review manuals have been the must have certification study aids used by those wishing to advance their careers with increased knowledge and certification This third edition of *Certified Ophthalmic Technician Exam Review Manual* provides the ultimate experience in exam preparation This best selling text is ideal for both individual and group study The explanatory answers contribute to your understanding of the material rather than only providing right or wrong feedback *Certified Ophthalmic Technician Exam Review Manual Third Edition* expands on what was originally the only study material available for this highly specialized exam It remains the go to source for the most questions and explanatory answers and has been updated to the latest IJCAHPO criteria With more than 1800 questions this text covers all 22 of IJCAHPO s criteria subjects at the COT level

Practical Management of Pediatric Ocular Disorders and Strabismus Elias Traboulsi, Virginia Utz, 2016-07-27 This case based approach to the management of pediatric eye diseases and strabismus teaches the novice as well as the experienced surgeon how to conceptualize and analyze surgical planning to maximize patient outcomes allowing readers to get inside the head of the surgeon Individual cases with clear instructions and an abundance of color illustrations

presented by experts in the field teach critical information one case at a time Practical Management of Pediatric Ocular Disorders and Strabismus provides the key guiding principles of a traditional textbook in an easy to digest format Each chapter of the text tackles a common or rare clinical situation and begins with the description of an illustrative case The chapters are structured with sections on case description differential diagnosis management and outcomes with a list of possible complications and the approach to their management *Equine Ophthalmology - E-Book* Brian Gilger,2010-10-07 With contributions from 18 of the world s leading equine experts Equine Ophthalmology provides the comprehensive and authoritative coverage you need to diagnose manage and operate on ocular disorders in horses Detailed illustrations facilitate the accurate identification of a wide range of ocular diseases while up to date and easily accessible clinical information helps you decide on the best course of treatment As the only comprehensive textbook on equine ophthalmology this reference provides the information you need to treat and manage equine ocular disease Seventeen expert contributors share their extensive insight while providing the most authoritative and current information available An extensive collection of high quality images helps you to identify and treat painful ocular diseases in horses Coverage of the financial impact of ocular disease on the equine industry provides a larger perspective on the importance of effective treatment Greater clinical detail concerning treatment diagnosis and procedures offers practical information for the general equine practitioner and an up to date resource for specialists Expert coverage of recent and far reaching advances in equine ophthalmology includes an expanded section on genetics A NEW chapter on Ophthalmologic Treatment in the Field written by expert Ann Dwyer offers an essential overview of ocular problems that require immediate attention NEW how to sections provide succinct directions on specific procedures for readers who are not well versed in surgery A detailed chapter outline starts each chapter and provides a quick overview of the chapter contents *Parsons' Diseases of the Eye* Sihota,Radhika Tandon,2023-11-04 Parsons Diseases of the Eye has served as a trusted textbook of Ophthalmology for undergraduate students for well over 100 years It also meets the needs of postgraduate students and practising clinicians as a useful guide to basic concepts and a comprehensive compendium of knowledge for ophthalmic care First published in 1907 the book soon became a fundamental text for students on account of its clear descriptive style and thorough coverage of the practical aspects of ocular disorders Over the years the book has maintained its clinical relevance through periodic revisions with suitable updates and hence retained its prime position at the top of its field The 19th edition of the book was a landmark adaptation to the context of the Indian subcontinent with a specially curated region specific content upgradation Subsequent editions including 24th current edition have served to build on this platform continuously evolving to match the changes in the academic curriculum remaining contemporary with global best practices and aligning with modern teaching learning philosophies NEW TO THIS EDITION Content thoroughly revised and updated Addition of new clinical pictures and digital ancillaries Information presentation re aligned for better understanding Key to competency codes added Guidelines for approach to investigations

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