

A review of ocular genetics and inherited eye diseases

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Received 23 April 2012; revised version accepted 2 November 2012

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-

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