



بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ



شبكة المعلومات الجامعية التوثيق الالكتروني والميكرو فيلم



شبكة المعلومات الجامعية

جامعة عين شمس

التوثيق الالكتروني والميكرو فيلم

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1992

**RECENT ADVANCES IN MOLECULAR
BIOLOGY AND GENETICS OF CORNEAL
DYSTROPHIES**

Essay

Submitted for partial fulfillment of M.Sc Degree in Ophthalmology

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(صام)

Abstract

Corneal dystrophies are known to be caused by mutations in genes encoding enzymes or structural proteins. These genes and their chromosomal location can be identified by recent molecular biology techniques. It is possible to develop cell cultures and animal models to study the function of relevant genes.

Gene therapy application in the field of corneal dystrophies is either through viral agents or physical agents. It is a promising treatment modality in the future.

Key Words:

Corneal dystrophies – gene therapy – virus – adenovirus – molecular biology.

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Introduction and Aim of the Work

Corneal dystrophies are known to be caused by mutations in genes encoding enzymes or structural proteins. During the last decade, advances in molecular genetics have disclosed new important information about these rare disorders. The specific genetic mutation and biochemical abnormalities responsible for corneal opacification and visual loss have been identified.

The aim of this work is to study the genetics and molecular biology of corneal dystrophies, and to examine the recent advances in the diagnosis, prophylaxis and therapy of these diseases.

The study will include a brief account on the molecular biology of corneal dystrophies. This is the means by which genetic information is transmitted from deoxyribonucleic acid (DNA) to ribonucleic acid (RNA) resulting in protein synthesis.

Gene therapy, either through genetic manipulations of donor cornea prior to transplantation or gene delivery to affected corneas, or limbal stem cell transplantation for corneal dystrophies of epithelial genesis, represents possible and promising therapies for corneal dystrophies in the future.

Some clinical application of genetics will also be included in the study. For example, carrier detection, presymptomatic diagnosis and population screening.

TYPES OF CORNEAL DYSTROPHIES

Introduction to corneal dystrophies:

Corneal dystrophies are a group of spontaneously appearing, mainly autosomal dominant, bilateral, stationary or slowly progressive corneal alterations. They develop in the absence of inflammation and systemic disease. Most present during the second decade.

Corneal dystrophies differ from corneal degenerations. The latter are usually age-related changes often accompanied by other ocular or systemic diseases. Corneal degenerations begin near the corneal periphery and lead to late visual impairment.