# LIPOID PROTEINOSIS

Study

Submitted For Partial Fulfillment of
The M.Sc. Degree In
**Dermatology and Venereology**

By

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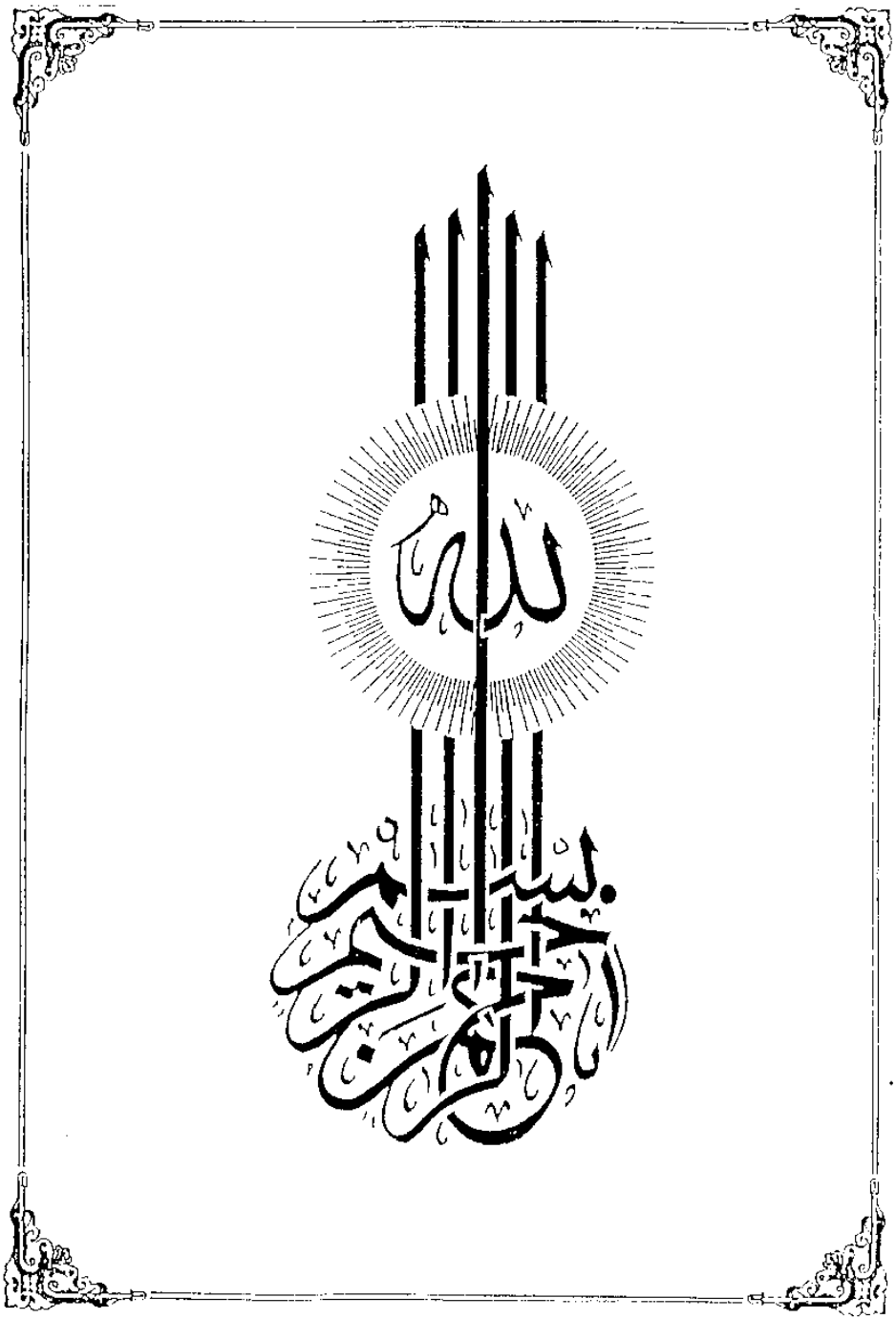

# Acknowledgment

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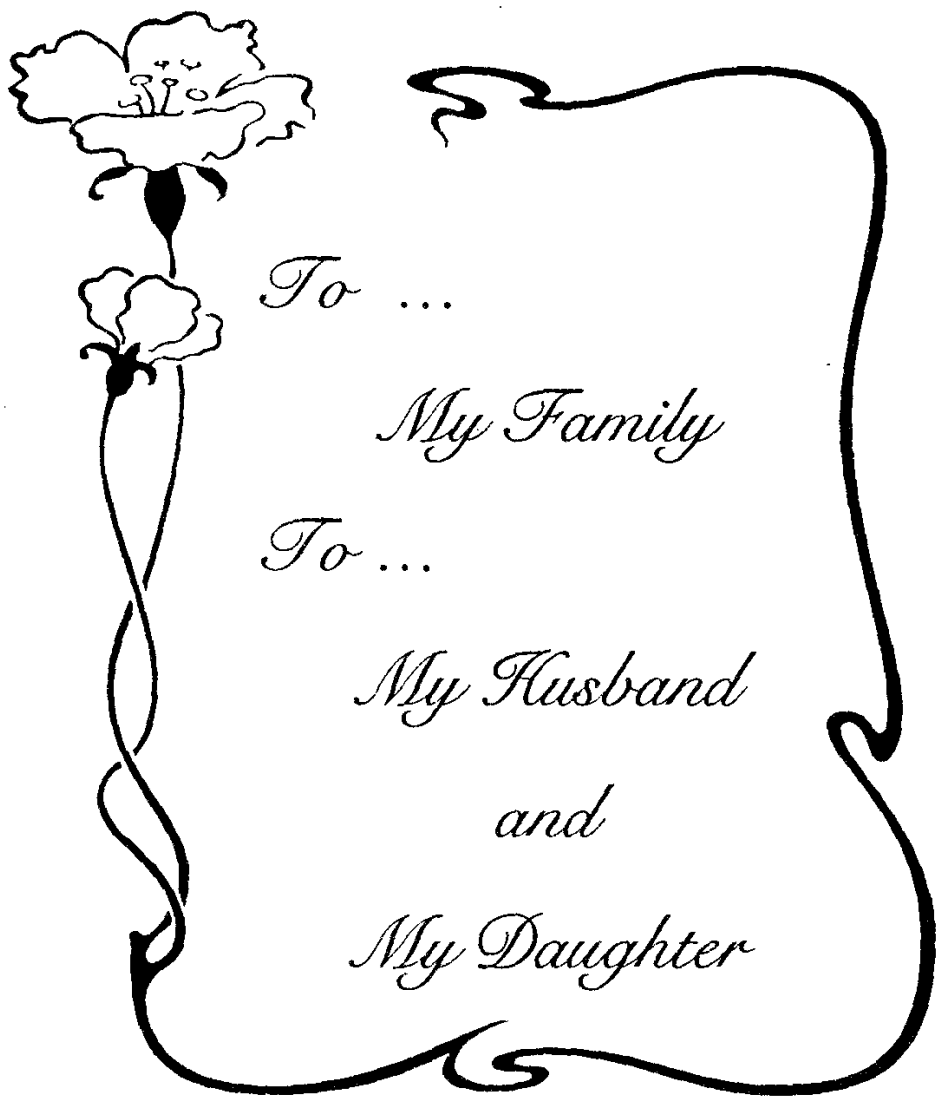
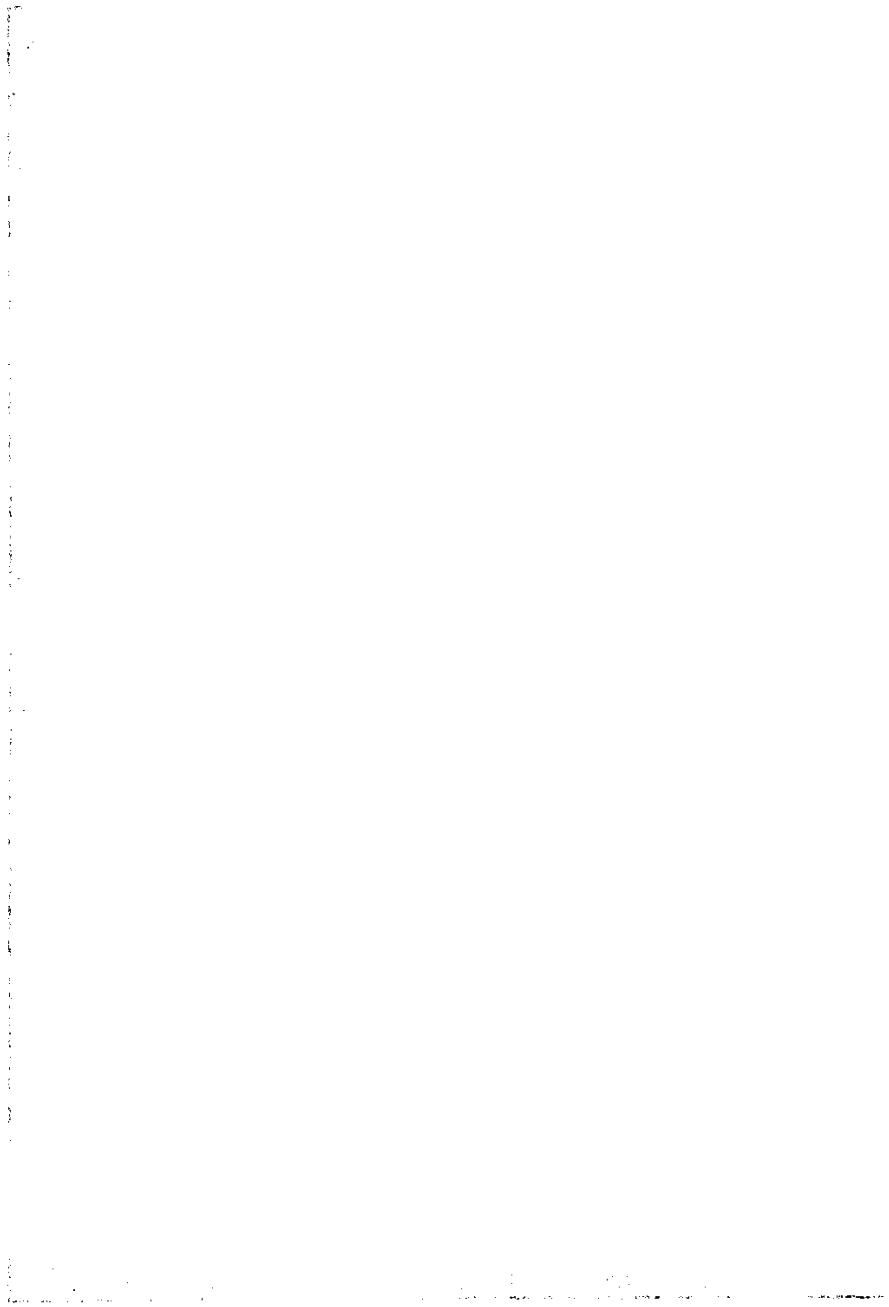
# Dedication



# CONTENTS



# LIST OF FIGURES

# *Introduction & Aim of The Study*

# INTRODUCTION

Lipoid proteinosis is a generalized hereditary disease with an autosomal recessive inheritance, affecting both sexes equally. First described by ***Urbach and Wiethe in (1929)*** as hyalinosis cutis et mucosae ***(Chaudhary and Daval, 1995)***. The disease is known by numerous names, including: Urbach-Wiethe diseae, lipoglycoproteinosis, lipoidosis cutis et mucosae, hyalinosis cutis et mucosae and lipoid proteinosis ***(Ramsey et al., 1985; Rizzo et al., 1997)***.

It is characterized by hyaline and lipid deposits in the skin and mucous membrane of the oral cavity, upper respiratory tract and some visceral organs ***(Chaudhary and Daval, 1995)***. It may also involves the central nervous system, gastrointestinal tract, lung, lymph nodes and striated muscles ***(Ozbek et al., 1994; Hafeez and Hussein, 1996)***.

The exact etiology of the disease is unknown, and several theories have been put forward to explain its possible mood of origin and pathogenesis.
It has been suggested that the causative factor may be an enzyme deficiency with a primary defect in the collagen metabolism that result in deposition of glycoproteins ***(Bauer et al., 1981; Harper et al., 1983)***.

Others suggested that the primary defect was degeneration of elastic and collagen fibres, with release of carbohydrates followed by a secondry deposition of lipoid material ***(Aziz et al., 1980)***.

Other theory suggested a mild form of serous inflammation caused by hypersensitivity to physiologic trauma. The terminal blood vessel of the skin and mucosa in patients suffering from lipoid proteinosis are congenitaly fragile. Exposure to some physiologic trauma may lead to the release of an enzyme from the vessel wall, causing increased permiability to circulating substances such as lipids and proteins. The released enzyme may also cause degeneration of the constituents of the perivascular connective tissues in the skin and mucous membrane ***(Aziz et al., 1980)***.

***Hashimoto*** in ***(1972)*** proposed that lipoid proteinosis may be the consequence of over production of basement membrane collagens by epithelial or endothelial cells and increased synthesis of extracellular glycoproteins by fibroblasts ***(Pierard et al., 1988; Chaudhary and Daval, 1995)***.

The most prominent and earliest symptom of lipoid proteinosis is hoarseness of voice since infancy as a result of involvement of the larynx and upper respiratory tract by hyaline deposits ***(Cinaz et al., 1993; Oezarmagan et la., 1993)***.