

Get Complimentary ebook access with the copy

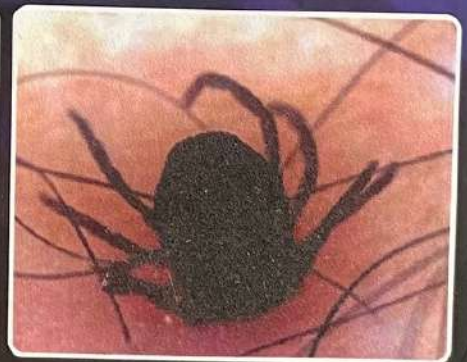
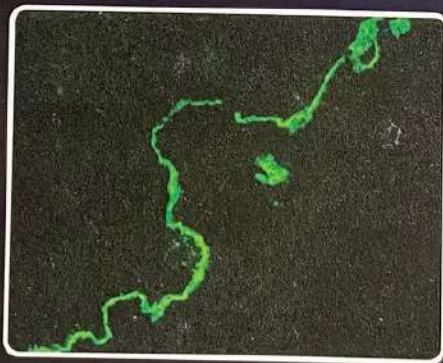
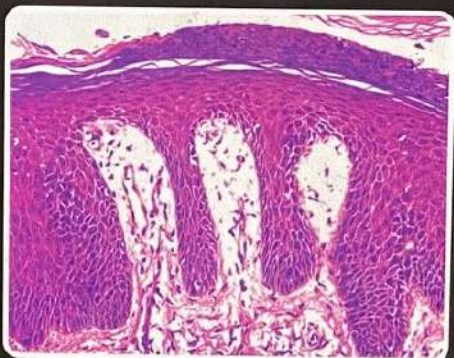
Log on bhalanidigital.com



VOLUME 1

IADVL Textbook of Dermatology

FIFTH EDITION



Editor-in-Chief

S. Sacchidanand

Co-Editors

Savitha A. S.

Shilpa K.

Shashi Kumar B. M.



BHALANI

Editors-in-Chief

S. Sacchidanand

MD, DVD, DHA, FRCP (Glasgow)
Honorary Vice Chancellor (2018–2021)
Rajiv Gandhi University of Health Sciences
Bengaluru, Karnataka

Co-Editors

Savitha A. S.

MD, DNB, FRGUHS
Professor
Department of Dermatology
Sapthagiri Institute of Medical
Sciences & Research Institute
Bengaluru
Karnataka

Shilpa K.

MD, FRGUHS
Associate Professor
Department of Dermatology
Bangalore Medical College and
Research Institute
Bengaluru
Karnataka

Shashi Kumar B. M.

MD, FIADVL
Associate Professor
Department of Dermatology
Mandya Institute of Medical
Sciences
Mandya
Karnataka

Associate Editor

Nagesh T. S.

Section Editors

Amina Asfiya Iqbal

Anupam Das

Archana Singal

Arun Inamadar

Avitus John Raakesh Prasad

B S Chandrashekar

Biju Vasudevan

Chander Grover

Deepika Pandhi

Dinesh Kumar D

Iffat Hassan

Jayadev Betkerur

Lakshmi D V

Madura C

Manish Gautam

Manjunath Shenoy

Nagesh T S

P Narasimha Rao

Ragunatha Shivanna

Raghunatha R Reddy

Rashmi Sarkar

Sahana M S

Savitha A S

Shashi Kumar B M

Shilpa K

Shital Poojary

Shivaswamy K N

Smitha Prabhu

Sujala S Aradhya

Umashankar N

Yogesh Marfatia

4	Immunology of Skin	<i>Brijesh Nair, Murlidhar Rajagopalan</i>	84
5	Principles of Clinical Diagnosis	<i>Archana Singal, Sidharth Sonthalia</i>	114
6	Basic Dermatopathology	<i>Uday Khopkar, Atul Dongre</i>	150
7	Basics of Dermoscopy	<i>Keshavmurthy A Adya, Ragunatha Shivanna</i>	187
8	Skin at Different Ages	<i>Shital Poojary, Saurabh Jaiswal</i>	195
9	Environment and Skin	<i>Wg Cdr Sanjiv Grover, Col Rajeshwari Dabas</i>	212

SECTION 2: PEDIATRIC DERMATOLOGY
Section Editors: Arun Inamadar, Sahana MS

10	Genodermatoses	<i>Aparna Palit, Arun C Inamadar</i>	237
11	Neonatal Skin Care and Skin Disorders	<i>Deepak Parikh, Sahana M Srinivas</i>	289
12	Nevi and other Developmental Defects	<i>Sangeeta Velaskar, Ragunatha Shivanna</i>	334
13	Ichthyosis and Ichthyosiform Disorders	<i>Neha Taneja, Vishal Gupta, Gomathy Sethuraman</i>	395
14	Disorders of Keratinization	<i>Timir Y Mehta, Neela Bhuptani, Pratik B Sheth</i>	429

SECTION 3: INFECTIONS AND INFESTATIONS
Section Editors: Manjunath Shenoy, Amina Asfiya Iqbal

15	Bacterial Infections	<i>Ramesh Bhat, Rochelle D Monteiro, Jyothi Jayaraman</i>	491
16	Cutaneous Tuberculosis	<i>Neetu Bhari, V Ramesh</i>	509
17	Cutaneous Nontuberculous Mycobacterial Infection	<i>Sivaranjini Ramassamy, Remya Raj R, Malathi Munisamy</i>	531
18	Superficial Fungal Infections	<i>Manjunath Shenoy</i>	537

Section

2

Pediatric Dermatology

SECTION EDITORS: ARUN INAMADAR,
SAHANA MS

10. Genodermatoses
11. Neonatal Skin Care and Skin Disorders
12. Nevi and other Developmental Defects
13. Ichthyosis and Ichthyosiform Disorders
14. Disorders of Keratinization

10

Genodermatoses

Aparna Palit • Arun C Inamadar

KEY MESSAGES

- Genodermatoses are inherited disorders commonly encountered in dermatology practice.
- The majority of these are life-long, involving multiple systems.
- Diagnosis of these group of disorders is mainly with clinical features, associations and genetic analysis.
- Treatment is multidisciplinary.
- Providing options for prenatal diagnosis to the parents and genetic counseling to prevent future occurrence is very important in these cases.

INTRODUCTION

Patients with genodermatoses constitute a significant section in dermatological practice. The majority of these are life-long, multisystem disorders. Some carry the risks of malignancy and premature death. Many genodermatoses cause cosmetic disfigurement, and patients present to the dermatologist for their unusual appearance. Management of these disorders is not only limited to the diagnosis of the particular patient but also probing into the family tree to know the inheritance pattern, providing options for prenatal diagnosis to their parents and genetic counseling to prevent further such occurrences. With this background, it is understandable that a basic knowledge of genetics is essential to dermatologists. The discussion in the following section includes the principles of genetic transmission of diseases and the terms related to it.

PRINCIPLES OF GENETIC TRANSMISSION

Transmission of characters through generations is determined by genes located on the chromosomes. Such transmission may or may not follow Mendelian laws. As a broad group, these are categorized as inherited disorders. A familial disorder is

recognized by the occurrence of a character in a family clearly in excess of its expected occurrence in the same population.¹ A congenital character is one present at or before birth.¹ Congenital and familial conditions do not necessarily imply genetic transmission. Non-Mendelian inherited disorders such as psoriasis and atopic dermatitis are quite common and show familial clustering frequently.²

The genotype of an individual indicates the characters transmitted through the genes. The physical expression of these characters is designated as phenotype. Autosomal characters are borne by autosomes (22 pairs of chromosomes) and sex-linked characters are carried by sex chromosomes (X/Y). Genes are located at particular chromosome loci as alleles. Two different alleles at a particular locus of a chromosome pair indicate heterozygosity, whereas identical alleles at a locus indicate homozygosity. Males with the expression of X-linked characters (unpaired alleles) are termed hemizygous.² A dominant character is defined as one whose phenotypic expression is possible in the heterozygous state. A recessive character manifests only in the homozygous state.

Four major patterns of genetic transmission are possible— autosomal dominant (AD), autosomal recessive (AR), X-linked dominant (X-L-D), and X-linked recessive (X-L-R). Sex-linked transmission may also occur through Y chromosomes (holandric transmission). The salient features of these are described in **Table 10.1**.^{1,2} A schematic representation of the different patterns of genetic transmission is shown in **Fig. 10.1**.

The above inheritance patterns are related to nuclear DNA. Disorders related to mitochondrial DNA show a maternal inheritance simulating X-linked inheritance patterns.³ Here, all sons and daughters of an affected woman are involved, but father-to-offspring transmission is not possible.²⁻⁴ Palmoplantar keratoderma with sensorineural deafness (mutation of *MTTS1*) follows this type of inheritance pattern.^{3,4} Nonspecific characters