

PLEASE READ—IMPORTANT INFORMATION

The *Gale Encyclopedia of Genetic Disorders* is a medical reference product designed to inform and educate readers about a wide variety of disorders, conditions, treatments, and diagnostic tests. Thomson Gale believes the product to be comprehensive, but not necessarily definitive. It is intended to supplement, not replace, consultation with a physician or other health care practitioner. While Thomson Gale has made substantial efforts to provide information that is accurate, comprehensive, and up-to-date, Thomson Gale makes no representations or warranties of any kind, including

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INTRODUCTION

The *Gale Encyclopedia of Genetic Disorders* is a unique and invaluable source for information regarding diseases and conditions of a genetic origin. This collection of over 430 entries provides in-depth coverage of disorders ranging from the exceedingly rare to the very well-known. In addition, several non-disorder entries have been included to facilitate understanding of common genetic concepts and practices such as Chromosomes, Genetic counseling, and Genetic testing.

This encyclopedia avoids medical jargon and uses language that laypersons can understand, while providing the thorough coverage of each disorder that medical professionals will also find beneficial. The *Gale Encyclopedia of Genetic Disorders* fills a gap between basic consumer health resources, such as single-volume family medical guides, and highly technical professional materials.

Each entry discussing a particular disorder follows a standardized format that provides information at a glance. The rubrics used are:

- Definition
- Description
- Genetic profile
- Demographics
- Signs and symptoms
- Diagnosis
- Treatment and management
- Prognosis
- Resources
- Key terms

INCLUSION CRITERIA

A preliminary list of diseases and disorders was compiled from a wide variety of sources, including professional medical guides and textbooks, as well as consumer guides and encyclopedias. The advisory board,

made up of seven medical and genetic experts, evaluated the topics and made suggestions for inclusion. Final selection of topics to include was made by the advisory board in conjunction with Thomson Gale editors.

ABOUT THE CONTRIBUTORS

The essays were compiled by experienced medical writers, primarily genetic counselors, physicians, and other health care professionals. The advisors reviewed the completed essays to insure they are appropriate, up-to-date, and medically accurate.

HOW TO USE THIS BOOK

The *Gale Encyclopedia of Genetic Disorders* has been designed with ready reference in mind.

- Straight **alphabetical arrangement** of topics allows users to locate information quickly.
- **Bold-faced terms** direct the reader to related articles.
- **Cross-references** placed throughout the encyclopedia point readers to where information on subjects without entries may be found.
- A list of **key terms** is provided where appropriate to define unfamiliar terms or concepts. Additional terms may be found in the **glossary** at the back of volume 2.
- The **Resources** section directs readers to additional sources of medical information on a topic.
- Valuable **contact information** for organizations and support groups is included with each entry. The appendix contains an extensive list of organizations arranged in alphabetical order.
- A comprehensive **general index** guides readers to all topics and persons mentioned in the text.

GRAPHICS

The *Gale Encyclopedia of Genetic Disorders* contains over 200 full-color illustrations, including photos and pedigree charts. A complete **symbol guide** for the

pedigree charts can be found at the beginning of each volume.

ACKNOWLEDGEMENTS

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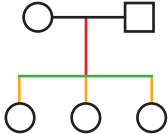

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SYMBOL GUIDE FOR PEDIGREE CHARTS

Pedigree charts are visual tools for documenting biological relationships in families and the presence of disorders. Using these charts, medical professionals such as geneticists and genetic counselors can analyze the genetic risk in a family for a particular trait or condition by tracking which individuals have the disorder and determining how it is inherited.

A standard set of symbols has been established for use in creating pedigree charts. Those found within the body of several entries in the encyclopedia follow the symbol guide explained on the next page. The exact style and amount of information presented on the chart varies for each family and depends on the trait or condition under investigation. Typically, only data that is directly related to the disorder being analyzed will be included. For more information, see the “Pedigree analysis” entry in the second volume.

Symbol Guide for Pedigree Charts

 Male	 Miscarriage
 Female	 Pregnancy terminated due to affected condition
 Affected male	 Elective termination of pregnancy
 Affected female	 Female with no children by choice
 Carrier male	 Female with no children due to medical infertility
 Carrier female	 Identical twin females
 Deceased male	 Fraternal twin females
 Deceased female	 Consanguineous relationship
 Male adopted into a family	 Relationship no longer exists
 Female adopted into a family	 Unknown family history
 Gender not specified	 Died at 79 years
 Pregnancy	 Diagnosed at 41 years
 Four males	
 Three females	Relationship line Line of descent Sibship line Individual line