

eReports

Rare Diseases e-Resource Guide

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Rare Diseases e-Resource Guide

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CONTENTS

INTRODUCTION

Definitions

Orphan Diseases and Drug Legislation

Healthcare Provider Knowledge and Awareness

Table 1-1: Summary of rare diseases electronic resources

COMPREHENSIVE REVIEW OF RARE DISEASES ELECTRONIC RESOURCES

National Organization for Rare Disorders

Genetic and Rare Diseases Information Center

Canadian Organization for Rare Disorders

Rare Diseases Europe

Orpha.net

Miscellaneous Resources

APPLICATION TO PRACTICE

Patient Case

Table 1-2: Porphyria patient case with e-resource information

SUMMARY

REFERENCES

ANNOTATED OUTSIDE RESOURCES

INTRODUCTION

Definitions

In the United States, a *rare (orphan) disease* is any disorder affecting fewer than 200,000 persons.^{1,2} Although each disease may be classified as "rare," when considered as a whole these diseases affect over 30 million Americans—three-fourths of them are children.¹⁻³ To put this into perspective, one in 10 Americans lives with a rare disease.⁴ In addition, there are nearly 7,000 rare diseases nationwide.^{2,3,5,6} *Orphan drugs* are medicinal products intended for the diagnosis, prevention, or treatment of life-threatening or very serious diseases or disorders that are classified as rare.⁷

Orphan Diseases and Drug Legislation

Historically, a product's expected sales would not recover the high-cost burden of bringing an orphan drug to market in a small population with a rare disorder.^{5,7} Thus, there was little incentive for pharmaceutical manufacturers to develop new products prior to the Orphan Drug Act of 1983. In fact, in the entire decade prior to 1983, only 10 new drugs indicated for rare diseases were brought to the market. After 1983, there was a 20-fold increase in drugs and biological products for rare diseases. Currently, there are more than