

Contents

Contributors	vii
Introduction	ix
Foreword	xi
Preface	xiii
Glossary of Molecular Genetics	xv
1. Principles of Molecular Genetics	1
<i>Jeffrey A. Towbin</i>	
2. Hereditary Amyloidosis	55
<i>Merrill D. Benson</i>	
3. Duchenne Muscular Dystrophy	73
<i>Ronald G. Worton and Elizabeth F. Gillard</i>	
4. Huntington's Disease	113
<i>James Gusella and Anne B. Young</i>	
5. HPRT Deficiency: Lesch-Nyhan Syndrome and Gouty Arthritis	129
<i>C. Thomas Caskey and Belinda J.F. Rossiter</i>	
6. Myotonic Dystrophy	147
<i>Allen D. Roses and Margaret A. Pericak-Vance</i>	
7. Neurofibromatosis	160
<i>Margaret R. Wallace and Francis S. Collins</i>	
8. Phenylketonuria	181
<i>Randy C. Eisensmith and Savio L.C. Woo</i>	2
9. Gilles de la Tourette Syndrome	199
<i>Michele C. LaBuda and David L. Pauls</i>	

10. The Tuberous Sclerosis Complex 215

M.R. Gomez and Moyra Smith

11. Charcot-Marie-Tooth Disease 233

Jeffery Vance and Tom Bird

12. Mitochondrial Myopathies 245

A.E. Harding and I.J. Holt

13. Fragile X Syndrome 263

David L. Nelson

Index 275