

# CONTENTS

## CONTRIBUTORS

### PREFACE

*Richard A. Gatti and Robert B. Painter*

## PART I. INTRODUCTION

### ATW5 PARTICIPANTS (Photo)

### BRIEF HISTORICAL OVERVIEW

*Robert P. Sedgwick . . . . . 1*

## PART II. ISOLATION OF A-T GENE(S)

### Cloning and characterization of a candidate gene for A-T complementation Group D

*Leon N. Kapp and John P. Murnane . . . . . 7*

### Precise localization of a gene responsible for ataxia- telangiectasia on chromosome 11q

*F. Cornelis, Michael James, D. Cherif, Takashi Tokino, J.  
Davies, D. Girault, C. Bernard, M.F. Croquette, D. Theau, H.  
Avet-Loiseau, Michael Litt, R. Berger, Yusuke Nakamura, Mark  
Lathrop and Cecille Julier . . . . . 23*

### How many A-T genes?

*Ethan Lange, Richard A. Gatti, Eric Sobel, Patrick Concannon  
and Kenneth Lange . . . . . 37*

### Isolation of human cDNAs that complement the ataxia- telangiectasia phenotype in cultured fibroblasts

*M. Stephen Meyn, Jennifer M. Lu-Kuo and Laura B.K.  
Herzing . . . . . 55*

### Complementation of the cellular A-T phenotype by gene transfer

*Yael Ziv, Tsafi Danieli, Galit Rotman, Adam Sartiel, Anat Bar-  
Shira, Nicholas G.J. Jaspers, Richard Swirski, Robert T.  
Schimke, Roger L. Eddy, Thomas B. Shows and Yosef  
Shiloh . . . . . 65*

### Use of microcell hybrids for analysis of the 11q23 region and improved localization of the A-T Group A/C genes

*Yusuke Ejima, Mitsuo Oshimura and Masao S. Sasaki . . . . . 75*

### AT-like radiosensitive rodent cell mutants: an alternative approach to the isolation of the A-T gene(s)

*Margaret Z. Zdzienicka, Gerald W C T Verhaegh, Wim Jongmans,  
Nicholas G.J. Jaspers, Mitsuo Oshimura, Michael R. James and  
Paul H.M. Lohman . . . . . 87*

## PART III. A-T HETEROZYGOTES AND COMPLEMENTATION

**Identification of A-T heterozygotes**

David Scott, Lucy A Jones, Sean A G Elyan, Ann Spreadborough,  
Richard Cowan and Gerald Ribiero . . . . . 101

**Correction of post-gamma ray DNA repair deficiency in ataxia-telangiectasia complementation group A fibroblasts by cocultivation with normal fibroblasts**

Malcolm C. Paterson and Razmik Mirzayans . . . . . 117

**The A-T gene does not make a major contribution to familial breast cancer**

Richard Wooster, Douglas F. Easton, Deborah Ford, Jonathan Mangion, Bruce A.J. Ponder, Julian Peto and Michael Stratton . . . . . 127

**Mammography screening for A-T heterozygotes**

Amos Norman and Rodney Withers . . . . . 137

## PART IV. DEFINING THE A-T DEFECT

**Lymphoid V(D)J recombination: accessibility and reaction fidelity in normal and ataxia-telangiectasia cells**

Chih-Lin Hsieh and Michael R. Lieber . . . . . 143

**Murine scid cells and human ataxia-telangiectasia cells complement each other's radiosensitivity**

Kenshi Komatsu . . . . . 155

**Ataxia-telangiectasia: defective in a p53-dependent signal transduction pathway**

Michael B. Kastan . . . . . 163

**DNA recombination in the transgenic mouse brain**

Linda Kingsbury and Itoshi Sakano . . . . . 175

## PART V. A-T VARIANTS

**Clinical variants of ataxia-telangiectasia**

Ozden Sanal, A. Izzet Berkel, Fugen Ersoy, Ilhan Tezcan and Haluk Topaloglu . . . . . 183

**Epidemiology of ataxia-telangiectasia in Italy**

Luciana Chessa and Massimo Fiorilli . . . . . 191

**Epidemiology of ataxia-telangiectasia in Costa Rica**

Oscar Porras, Olga Arguendas, Margarita Arata, Max Barrantes, Luis Gonzalez and Elizabeth Saenz . . . . . 199

**Clinical and cellular heterogeneity in ataxia-telangiectasia**

A. Malcolm R. Taylor, Carmel M. McConville and C. Geoffrey Woods, Phillip J. Byrd and Diana Hernandez . . . . . 209

**PART VI. OVERVIEWS**

|                                                     |         |
|-----------------------------------------------------|---------|
| <b>Biochemical defects in ataxia-telangiectasia</b> |         |
| <i>Martin F. Lavin</i> . . . . .                    | 235     |
| <b>Radiobiology of ataxia-telangiectasia</b>        |         |
| <i>Robert B. Painter</i> . . . . .                  | 257     |
| <b>Treatment of ataxia-telangiectasia</b>           |         |
| <i>Susan L. Perlman</i> . . . . .                   | 269     |
| <br><b>INDEX</b> . . . . .                          | <br>279 |