

# Table of Contents

## MALFORMATIONS

### Forebrain Patterning Defects

- 1. Neural Tube Defects 2
- 2. Midline Patterning Defects 14
- 3. Microcephaly 26

### Cell Migration and Specification Disorders

- 4. Lissencephaly, Type I 34
- 5. Lissencephaly, Type II (Cobblestone) 42
- 6. Polymicrogyria 49
- 7. Cerebral Heterotopia 52
- 8.1. Epilepsy Part I: Cortical Dysplasia 61
- 8.2. Epilepsy Part I: Hemimegalencephaly 67
- 9.1. Epilepsy Part II: Rasmussen Syndrome, Hippocampal Sclerosis, Granule Cell Dysplasia 73
- 9.2. Tuberous Sclerosis 79

### Cerebellum, Hindbrain, and Spinal Patterning Defects

- 10. Chiari Malformations 90
- 11. Dandy Walker Malformation 95
- 12. Cerebellar Heterotopia and Dysplasia 100
- 13. Brainstem Malformations 105
- 14. Spinal Cord Lesions 109

### Anomalies of Cerebral Vasculature

- 15. Pediatric Vascular Malformations 116

### Hydrocephalus

- 16. Hydrocephalus 126

### Chromosomal Changes

- 17. CNS Manifestations of Chromosomal Change 132

## SECONDARY MALFORMATIONS AND DESTRUCTIVE PATHOLOGIES

### Disruptions of Development

- 18. Antenatal Disruptive Lesions 144

### Specific Destructive Disorders

- 19. Hemorrhagic Lesions 150
- 20. White Matter Lesions in the Perinatal Period 156
- 21. Grey Matter Lesions 171
- 22. Acquired Vascular Lesions in Children 176
- 23. Pediatric Head Injury 184

### SIDS

- 24. Sudden Infant Death Syndrome 194

## METABOLIC AND EXOGENOUS TOXINS

- 25. Kernicterus 206
- 26. Lesions Induced by Toxins 209

## METABOLIC DISORDERS

- 27.1 Disorders of Carbohydrate Metabolism. Introduction and Lysosomal Disorders 214
- 27.2 Disorders of Carbohydrate Metabolism. Polyglucosan Disorders. 221
- 27.3. Disorders of Carbohydrate Metabolism. The Congenital Disorders of Glycosylation. 226
  - 28.1. Sphingolipidoses 235
  - 28.2. GM1 Gangliosidosis 237
  - 28.3. GM2 Gangliosidosis 240
  - 28.4. Niemann-Pick Disease Types A and B 244
  - 28.5. Gaucher Disease 247
  - 28.6. Farber Disease 251
  - 28.7. Fabry Disease 253
  - 28.8. Metachromatic Leukodystrophy 256
  - 28.9. Multiple Sulfatase Deficiency 261
  - 28.10. Globoid Cell Leukodystrophy (Krabbe disease) 263
  - 28.11. Sphingolipid Activator Protein Deficiency 266
- 29.1 The Neuronal Ceroid Lipofuscinoses 270
- 29.2. Palmitoyl-protein Thioesterase 1 Deficiency with Granular Osmiophilic Deposits (CLN1) 271
- 29.3. Classic Late-infantile NCL with Tripeptidyl-Peptidase I Deficiency (CLN2) 274
- 29.4. Juvenile NCL with Mutations in CLN3 Gene (CLN3) 277
- 29.5. Rare Forms of Neuronal Ceroid Lipofuscinoses 280
- 30. Niemann-Pick Type C Disease 283
- 31. Peroxisomal Disorders 287
- 32. Mitochondrial Disorders 296
- 33. Disorders of Amino Acid Metabolism 303
- 34. Pelizaeus-Merzbacher Disease 311
- 35. Cockayne Syndrome 318
- 36. Vanishing White Matter Disease 325
- 37. Alexander Disease 331
  - 38.1. Infantile Neuroaxonal Dystrophy (Seitelberger Disease) 337
  - 38.2. Neurodegeneration with Brain Iron Accumulation Type 1 340
- 39. Spinal Muscular Atrophy 344
- 40. Neuropathology of Autism 349
- 41. Rett Syndrome 353

## INFECTIOUS DISEASES

- 42. Intrauterine Infections 360
- 43. Perinatal and Postnatal Infections 367

## CONTRIBUTORS

380

## INDEX

383