
Contents

1 The Apparently Blind Infant	1
Introduction	1
Neuro-Ophthalmologic Clues	1
Crossmodality of Sensory Input	4
Assessment of Vision	5
Hereditary Retinal Disorders	7
Leber Congenital Amaurosis	7
Ciliopathies	10
Congenital Stationary Night Blindness	12
Achromatopsia	12
Congenital Optic Nerve Disorders	13
Cortical Visual Insufficiency	13
Causes of Cortical Visual Insufficiency	15
Associated Neurologic and Systemic Disorders	25
Characteristics of Visual Function	27
Neuro-Ophthalmologic Findings	28
Retrograde Transsynaptic Degeneration	30
Diagnostic and Prognostic Considerations	31
Role of Visual Attention	33
Subcortical Visual Loss (Periventricular Leukomalacia)	34
Neuroimaging Abnormalities and Their Implications	34
Neuro-Ophthalmologic Findings	36
Perceptual Difficulties	38
Dorsal and Ventral Stream Dysfunction	40
Pathophysiology	42
Intraventricular Hemorrhage	43
Periventricular and Intraventricular Hemorrhage	43
Hemianopic Visual Field Defects in Children	44
Causes	45
Homonymous Hemiopic Atrophy	46
Afferent Pupillary Defect	46
Adaptive Strategies	46
Nonadaptive Strategies	46
Delayed Visual Maturation	47
Clinical Profile	47
Neurodevelopmental Aspects	47
Electrophysiologic Correlates	48
Classification	48
Overlap with Amblyopia	49
Optic Disc Appearance	49
Neuroimaging	49
Differential Diagnosis	50

Blindsight.....	51
The Effect of Total Blindness on Circadian Regulation	53
Horizons.....	56
References.....	57
2 Congenital Optic Nerve Anomalies	75
Introduction.....	75
Optic Nerve Hypoplasia.....	75
Background.....	75
Ophthalmoscopic Appearance	76
Histopathology	76
Visual Function.....	76
Measurement Techniques.....	78
Hypoplasia vs. Apoptosis.....	78
Hypopituitarism	78
Risk of Sudden Death	79
MR Imaging Abnormalities	80
Segmental Optic Nerve Hypoplasia.....	82
Pathogenesis.....	84
Genetics.....	84
Excavated Optic Disc Anomalies.....	85
Morning Glory Disc Anomaly (the Moyamoya Optic Disc)	85
Moyamoya Disease	88
PHACE Syndrome	88
Miscellaneous Disorders.....	88
Associated Retinal Detachment	88
Optic Disc Coloboma.....	90
Terminology.....	90
Ophthalmoscopic appearance	90
Visual Function.....	93
Genetics	93
Associated Findings.....	93
Associated Retinal Detachment	94
Nosology	94
Peripapillary Staphyloma.....	95
Megalopapilla.....	96
Optic Pit	97
Papillorenal Syndrome (The Vacant Optic Disc).....	99
Congenital Tilted Disc Syndrome.....	100
Optic Disc Dysplasia.....	101
Congenital Optic Disc Pigmentation	101
Aicardi Syndrome	103
Doubling of the Optic Disc.....	105
Optic Nerve Aplasia.....	106
Myelinated (Medullated) Nerve Fibers.....	109
Albinotic Optic Disc	110
References.....	111
3 The Swollen Optic Disc in Children	121
Introduction.....	121
Papilledema.....	122
Ophthalmoscopic Appearance	122
Papilledema in Infancy.....	122
Pathophysiology.....	123

Neurological Symptoms and Signs	123
Idiopathic Intracranial Hypertension (IIH) in Children	125
Major Neurologic Causes of Papilledema	136
Optic Disc Swelling Secondary to Systemic Disease	137
Diabetic Papillopathy	137
Malignant Hypertension	138
Sarcoidosis	139
Leukemia	140
Cystinosis	141
Congenital Cyanotic Heart Disease	141
Craniosynostosis Syndromes	142
Nonaccidental Trauma (Shaken Baby Syndrome)	144
Neurocysticercosis	145
Mucopolysaccharidosis	147
Infantile Malignant Osteopetrosis	147
Malaria	147
Paraneoplastic	147
Optic Disc Swelling Due to Ocular Disease	148
Uveitis	148
Posttraumatic Optic Disc Swelling	149
Intrinsic Optic Disc Tumors	150
Optic Disc Hemangioma	150
Tuberous Sclerosis	150
Optic Disc Glioma	151
Combined Hamartoma of the Retina and RPE	151
Retrobulbar Tumors	151
Optic Neuritis in Children	152
General History and Physical Examination	153
Differential Diagnosis	153
Temporal Course	160
Systemic Evaluation	160
Prognosis	161
Treatment	161
Neuroretinitis	162
Leber Idiopathic Stellate Neuroretinitis	162
IRVAN	165
Ischemic Optic Neuropathy	166
Autoimmune Optic Neuropathy	166
Pseudopapilledema	167
Optic Disc Drusen	167
Epidemiology	168
Ophthalmoscopic Appearance in Children	168
Clinical Overlap with Papilledema	168
Ophthalmoscopic Distinction from Papilledema	168
Fluorescein Angiographic Appearance	170
Ancillary Studies	171
Histopathology	172
Pathogenesis	172
Ocular Complications	173
Ocular and Systemic Associations	176
Local Causes of Pseudopapilledema	180
Natural History and Prognosis	180
References	181

4	Optic Atrophy in Children	199
	Introduction.....	199
	Epidemiology	202
	Optic Atrophy Associated with Retinal Disease.....	204
	Congenital Optic Atrophy Versus Hypoplasia.....	205
	Causes of Optic Atrophy in Children.....	205
	Compressive/Infiltrative Intracranial Lesions	205
	Noncompressive Causes of Optic Atrophy in Children with Brain Tumors.....	219
	Hereditary Optic Atrophy	221
	Toxic/Nutritional Optic Neuropathy	236
	Neurodegenerative Disorders.....	238
	Optic Atrophy Due to Hypoxia-Ischemia	247
	Traumatic Optic Atrophy	248
	Miscellaneous Causes	249
	Summary of the General Approach to the Child with Optic Atrophy	250
	References.....	251
5	Transient, Unexplained, and Psychogenic Visual Loss in Children	275
	Introduction.....	275
	Transient Visual Loss.....	276
	Migraine	276
	Epilepsy.....	287
	Posttraumatic Transient Cerebral Blindness	292
	Cardiogenic Embolism.....	292
	Nonmigrainous Cerebrovascular Disease	293
	Miscellaneous Transient Visual Disturbances in Children	293
	Summary of Clinical Approaches to the Child with Transient Visual Disturbances	298
	Laboratory Evaluation of Transient Visual Disturbances in Children	300
	Unexplained Visual Loss in Children	300
	Causes of Unexplained Visual Loss in Childhood.....	300
	Psychogenic Visual Loss in Children	306
	Clinical Profile	306
	Neuro-ophthalmologic Findings	307
	Categories of Psychogenic Visual Loss in Children	308
	Management of Psychogenic Visual Loss in Children	309
	When to Refer Children with Psychogenic Visual Loss for Psychiatric Treatment	310
	Horizons.....	311
	References.....	311
6	Ocular Motor Nerve Palsies in Children	325
	Introduction.....	325
	Relevant History.....	325
	Physical Examination.....	326
	Neuroimaging	328
	Oculomotor Nerve Palsy	328
	Anatomy.....	328
	Clinical Features	329
	Partial Forms of Oculomotor Palsy.....	331
	Oculomotor Synkinesis.....	333
	Causes of Third Nerve Palsy.....	334
	Differential Diagnosis	340
	Management.....	342

Trochlear Nerve Palsy.....	343
Anatomy.....	343
Neuroimaging.....	344
Clinical Features.....	345
Bilateral Trochlear Nerve Palsy.....	348
Causes of Trochlear Nerve Palsy.....	349
Differential Diagnosis.....	353
Treatment.....	355
Abducens Nerve Palsy.....	357
Anatomy.....	357
Clinical Features.....	357
Causes of Sixth Nerve Palsy.....	358
Differential Diagnosis.....	362
Duane Retraction Syndrome.....	362
Systemic Associations.....	368
Multiple Cranial Nerve Palsies in Children.....	374
Horizons.....	374
References.....	375
7 Complex Ocular Motility Disorders in Children.....	393
Introduction.....	393
Strabismus in Children with Neurological Dysfunction.....	394
Cerebral Palsy.....	394
Craniosynostosis.....	394
Cerebellar Disorders.....	396
Visuovestibular Disorders.....	398
Neurologic Esotropia.....	399
Neurologic Exotropia.....	402
Convergence Insufficiency.....	402
Accommodative Paresis.....	403
Skew Deviation.....	404
Gaze Palsies, Gaze Deviations, and Ophthalmoplegia.....	407
Horizontal Gaze Palsy.....	407
Vertical Gaze Palsies in Children.....	414
Diffuse Ophthalmoplegia in Children.....	417
Transient Ocular Motor Disturbances of Infancy.....	434
Transient Neonatal Strabismus.....	434
Transient Idiopathic Nystagmus.....	435
Tonic Downgaze.....	435
Tonic Upgaze.....	437
Neonatal Opsoclonus.....	439
Transient Vertical Strabismus in Infancy.....	439
Congenital Cranial Dysinnervation Syndromes.....	439
Congenital Ptosis.....	439
Marcus Gunn Jaw Winking (Trigemino-Oculomotor Synkinesis).....	440
Congenital Fibrosis Syndrome.....	441
Congenital Horizontal Gaze Palsy with Scoliosis.....	444
Möbius Sequence.....	445
HOXA1 Mutations.....	446
Monocular Elevation Deficiency, or “Double Elevator Palsy”.....	447
Brown Syndrome.....	450
Other Pathologic Synkineses.....	452
Internuclear Ophthalmoplegia.....	452

Clinical Signs	452
Pathophysiology	453
Clinical Variants	453
Causes	453
Treatment	454
Cyclic, Periodic, or Aperiodic Disorders Affecting Ocular Structures	454
Ocular Neuromyotonia	458
Ocular Motor Adaptations and Disorders in Patients with Hemispheric Abnormalities	458
Eyelid Abnormalities in Children	458
Eye Movement Tics	458
Excessive Blinking in Children	459
Hemifacial Spasm	461
Congenital Ptosis	462
Eyelid Retraction	463
Apraxia of Eyelid Opening	463
Pupillary Abnormalities	464
Congenital Bilateral Mydriasis	464
Adie Syndrome	466
Horner Syndrome	467
References	470
8 Nystagmus in Children	495
Introduction	495
Infantile Nystagmus	496
Clinical Features	496
Onset	497
Terminology	498
History and Physical Examination	498
Diagnostic Testing	500
Overlap with Strabismus	502
Eye Movement Recordings	502
Ocular Stabilization Systems	502
Suppression of Oscillopsia	505
Contrast Sensitivity and Pattern Detection Thresholds in Infantile Nystagmus	506
Theories of Causation	507
Genetics	508
Associated Visual Disorders	509
Neuroimaging	522
Treatment	523
Spasmus Nutans	529
Russell Diencephalic Syndrome of Infancy	532
Monocular Nystagmus	533
Nystagmus Associated with Infantile Esotropia	533
Torsional Nystagmus	533
Horizontal Nystagmus	533
Latent Nystagmus	533
Treatment	536
Nystagmus Blockage Syndrome	537
Treatment	537
Vertical Nystagmus	537
Upbeating Nystagmus in Infancy	537

Congenital Downbeat Nystagmus.....	538
Acquired Downbeat Nystagmus	539
Hereditary Vertical Nystagmus	539
Periodic Alternating Nystagmus	540
Seesaw Nystagmus.....	540
Congenital versus Acquired Seesaw Nystagmus	541
Saccadic Oscillations that Simulate Nystagmus	542
Convergence-Retraction Nystagmus.....	542
Opsoclonus and Ocular Flutter	542
Voluntary Nystagmus.....	544
Ocular Bobbing.....	545
Neurological Nystagmus.....	545
Leigh Subacute Necrotizing Encephalomyelopathy	546
Pelizaeus-Merzbacher Disease.....	546
Joubert Syndrome	546
Santavuori-Haltia Disease.....	547
Infantile Neuroaxonal Dystrophy.....	547
Carbohydrate-Deficient Glycoprotein Syndromes.....	547
Down Syndrome	547
Hypothyroidism	547
Maple Syrup Urine Disease	547
Lysosomal Disorders.....	548
Nutritional Nystagmus	548
Epileptic Nystagmus	548
Cobalamin C Methylmalonic Aciduria and Homocystinuria	548
Familial Vestibulocerebellar Disorder	548
Vestibular Paroxysmia.....	548
Summary.....	548
References.....	552
9 Torticollis and Head Oscillations.....	569
Introduction.....	569
Torticollis	569
Ocular Torticollis	570
Refractive Causes of Torticollis	570
Head Tilts	571
Head Turns.....	581
Vertical Head Positions	584
Head Oscillations	585
Head Oscillations with Nystagmus.....	586
Head Oscillations Without Nystagmus	588
References.....	590
10 Neuro-Ophthalmologic Manifestations of Neurodegenerative Disease in Childhood.....	597
Introduction.....	597
Neuronal Disease	601
Neuronal Ceroid Lipofuscinosis	601
Lysosomal Diseases	603
Gangliosidoses	604
Mucopolysaccharidoses	607
Subacute Sclerosing Panencephalitis.....	610
Leukoencephalopathies.....	610
Metachromatic Leukodystrophy	612

Canavan Disease (Spongy Degeneration of Cerebral White Matter)	613
Krabbe Disease	614
Pelizaeus–Merzbacher Disease	614
Cockayne Syndrome	616
Alexander Disease.....	617
Spastic Ataxia of Charlevoix-Saguenay.....	618
Sjögren–Larsson Syndrome	618
Kjellin Syndrome	618
Cerebrotendinous Xanthomatosis	619
Peroxisomal Disorders	620
Zellweger Spectrum	620
X-Linked Adrenoleukodystrophy	621
Neonatal Leukodystrophy	624
Basal Ganglia Disease.....	624
Pantothenate Kinase-Associated Neurodegeneration (PKAN).....	624
Disorders of Copper Metabolism.....	625
Menkes Disease	625
Wilson Disease.....	626
Autosomal Recessive Cataracts, Hearing Loss, and Neurodegeneration	627
Aminoacidopathies and Other Biochemical Defects	627
Maple Syrup Urine Disease	627
Homocystinuria.....	627
Abetalipoproteinemia.....	628
Mitochondrial Encephalomyelopathies	629
Chronic Progressive External Ophthalmoplegia (CPEO).....	630
Leigh Subacute Necrotizing Encephalomyelopathy	632
Mitochondrial Encephalomyelopathy and Stroke-Like Episodes (MELAS)	634
Myoclonic Epilepsy and Ragged Red Fibers (MERRF).....	634
POLG and Twinkle Mutations	634
Mitochondrial Depletion Syndrome	635
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD).....	635
Congenital Disorders of Glycosylation.....	636
Horizons.....	636
References.....	637
11 Neuro-Ophthalmologic Manifestations of Systemic and Intracranial Disease ..	649
Introduction.....	649
The Phakomatoses	649
Neurofibromatosis-1	649
Neurofibromatosis 2.....	660
Tuberous Sclerosis	664
Sturge-Weber Syndrome	668
von Hippel–Lindau Disease.....	671
Ataxia-Telangiectasia.....	673
Linear Nevus Sebaceous Syndrome.....	675
Klippel–Trenaunay–Weber Syndrome.....	678
Brain Tumors	681
Suprasellar Tumors	681
Arachnoid Cysts.....	682
Cavernous Sinus Lesions	683
Hemispheric Tumors.....	683
Posterior Fossa Tumors.....	684
Tumors of the Pineal Region.....	692
Meningiomas.....	694

Epidermoids and Dermoids.....	694
Gliomatosis Cerebri	694
Metastasis	695
Complications of Treatment of Intracranial Tumors in Children	695
Hydrocephalus	696
Hydrocephalus Due to CSF Overproduction	697
Noncommunicating Hydrocephalus.....	697
Communicating Hydrocephalus.....	698
Common Causes of Hydrocephalus in Children.....	698
Clinical Features of Hydrocephalus.....	707
Effects and Complications of Treatment.....	710
Vascular Lesions	713
AVMs	713
Cavernous Angiomas	716
Intracranial Aneurysms.....	716
Internal Carotid Artery Agenesis, Aplasia, and Hypoplasia.....	717
Isolated Venous Ectasia.....	717
Fistulas	717
Craniocervical Arterial Dissection.....	717
Strokes in Children	717
Cerebral Venous Thrombosis.....	720
Cerebral Dysgenesis and Intracranial Malformations.....	721
Destructive Brain Lesions	723
Malformations Due to Abnormal Stem Cell Proliferation or Apoptosis	725
Malformations Due to Abnormal Neuronal Migration.....	727
Malformations Secondary to Abnormal Cortical Organization and Late Migration	729
Anomalies of the Hypothalamic–Pituitary Axis	733
Encephaloceles.....	734
Cerebellar Malformations	737
Miscellaneous Multisystem Neuro-Ophthalmologic Disorders.....	737
Congenital Corneal Anesthesia.....	741
Posterior Reversible Encephalopathy Syndrome.....	741
Cerebroretinal Vasculopathies	742
Proteus Syndrome	742
PHACE Syndrome	742
Goldenhar Syndrome (Oculoauriculovertebral Dysplasia).....	743
Wildervanck (Cervico-oculo-acoustic) Syndrome.....	743
Delleman (Oculocerebrocutaneous) Syndrome	744
Encephalocraniocutaneous Lipomatosis	745
Incontinentia Pigmenti (Bloch-Sulzberger Syndrome).....	745
References.....	746
Index.....	777



<http://www.springer.com/978-1-4939-3382-2>

Pediatric Neuro-Ophthalmology

Brodsky, M.C.

2016, XXI, 823 p. 294 illus., 146 illus. in color.,

Hardcover

ISBN: 978-1-4939-3382-2