

---

## Contents

<b>1 The Apparently Blind Infant</b>	<b>1</b>
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 Hemioptic 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
<b>2 Congenital Optic Nerve Anomalies .....</b>	<b>75</b>
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
<b>3 The Swollen Optic Disc in Children .....</b>	<b>121</b>
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

<b>4 Optic Atrophy in Children .....</b>	<b>199</b>
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
<b>5 Transient, Unexplained, and Psychogenic Visual Loss in Children .....</b>	<b>275</b>
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
<b>6 Ocular Motor Nerve Palsies in Children .....</b>	<b>325</b>
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
<b>7 Complex Ocular Motility Disorders in Children .....</b>	<b>393</b>
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
<b>8 Nystagmus in Children .....</b>	<b>495</b>
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
<b>9 Torticollis and Head Oscillations.....</b>	<b>569</b>
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
<b>10 Neuro-Ophthalmologic Manifestations of Neurodegenerative Disease in Childhood.....</b>	<b>597</b>
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
<b>11 Neuro-Ophthalmologic Manifestations of Systemic and Intracranial Disease ..</b>	<b>649</b>
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
<b>Index.....</b>	<b>777</b>

Pediatric Neuro-Ophthalmology

Brodsky, M.C.

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

Hardcover

ISBN: 978-1-4939-3382-2