Congenital Anomalies of the Optic Disc

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Foreword

The subject of congenital anomalies of the optic disc is a difficult one, but it is masterfully treated by the authors. Their presentation is complete yet concise, interesting, and of great practical value. The authors' thorough survey of the literature has produced a mine of useful information for both the clinician and the pathologist. What makes reading through the book both enjoyable and easy is the happy balance among clinical, anatomic, embryologic, and genetic considerations. When applicable, a number of therapeutic approaches are discussed. The illustrations are profuse, well adapted to the subject, and of outstanding quality.

This book will become required reading for all those with an interest in changes of the optic disc—whether clinician or pathologist, ophthalmologist or neurologist. The authors have filled a great need in the literature about the human fundus; the scientific community will be grateful for their enormous and successful effort in producing an outstanding publication.

Charles L. Schepens, M.D.
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Preface

Over the past several years it has become apparent to us that a concise clinical work dealing with congenital anomalies of the optic disc was lacking. Because of our particular interest in children, we believed it was important to pursue the gathering of information to help the clinician recognize and manage these entities. While certain of the abnormalities described herein are not congenital, all can be seen in children and are important in the differential diagnosis of congenital optic disc anomalies. We hope that this book will help to further disseminate the knowledge obtained by the many talented physicians whose previous efforts have made such work possible.

The production of any book requires more than simply the efforts of the authors. We are indebted to the many individuals who have helped to make this publication possible and would like to acknowledge them as a small measure of our thanks. Gwendolyn Pearson spent countless hours typing and retyping the manuscript, an effort made possible through the generosity of the Research Department of Wills Eye Hospital. Jo Hoffman and Angeles Roca devoted a great deal of time to preparing the photographs. Most of the artwork was drawn by Laurel Cook, while Gayle Wallace also helped. Gloria Lewis and Fleur Weinberg deserve much credit for excellent detective work in tracking down the older and more obscure references. The fundus photographs were taken by Jamie Nicholl, Dennis Orlack, Richard Lambert, and Terry Tomer. We are grateful to Drs. William Annesley, Douglas Anderson, James Augsburger, Robert Behar, William Benson, Joseph Calhoun, Larry Donoso, Stephen Felton, Richard Goldberg, W. Richard Green, Sohan Hayreh, Alfred Lucier, Larry Magargal, Peter Savino, Norman Schatz, Henry Scimeca, Jerry Shields, G. Webster Taggart, Paul Whitmore, Myron Yanoff, and Lorenz Zimmerman for allowing their photographs to be reproduced. Last, but by no means least, we would like to thank our families for their patience and constant support throughout this whole endeavor.

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OPHTHALMOSCOPIC

The optic disc (or optic nerve head or papilla) normally is seen opthalmoscopically as a round or slightly ovoid yellow-pink structure (Fig. 1-1) with its center located approximately 4 mm superonasal to the foveola. In those eyes where the vertical and horizontal diameters are asymmetrical, the vertical dimension is usually the slightly longer of the two. The average nerve head is usually referred to

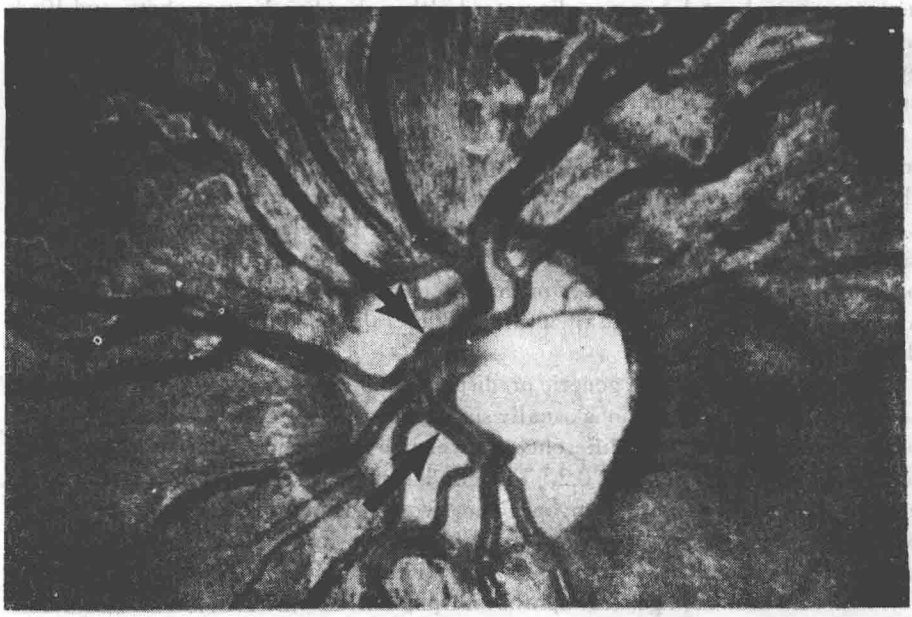

Fig. 1-1. Normal optic nerve head. The papilla is ovoid and has a slightly longer vertical diameter than horizontal. The retinal arteries (arrows) enter nasal to the point of exit of the retinal veins.

Table 1-1
Incidence of Refractive Errors in 500 Unselected Patients From a General Ophthalmic Practice

Refractive Error in Diopters	Percentage of Patients	
> +5.00	1	
+3.00 to +5.00	3	24
+1.00 to +3.00	20	
−1.00 to +1.00	53	
−3.00 to −1.00	13	
−5.00 to −3.00	7	23
> −5.00	3	

Data from Snydacker.[8]
Cylinders are compensated for by conversion to the spherical equivalent.

as measuring about 1.5 mm in diameter,[1] although when Franceschetti and Bock averaged the horizontal and vertical diameters of the nerve head in 100 normal eyes they found a mean of 1.62 mm.[2] A distribution of optic disc diameters, with standard deviations from the mean, is shown in Fig. 1-2.

Usually, a physiologic cup is present centrally on the optic disc, and while it may range from less than 0.1 to 0.9 of the disc diameter, cupping of 0.8 or greater of the horizontal disc diameter is seen in less than 5 percent of normal eyes (Fig. 1-3).[3,4] It should be noted that there is a positive correlation between larger cup size and increased depth of the cup.[3] With larger cups, the retinal vessels tend to pass horizontally along the floor of the cup until they curve anteriorly to proceed along the sides of the cup and then turn 90° laterally to course on the surface of the disc.[3]

There appears to be a genetic predisposition of the cup/disc (C/D) ratio, and in the same patient the ratio is usually similar for the two eyes.[5] In about 8 percent of normal individuals the difference between the two cups exceeds 0.1, while in only 1 percent does it exceed 0.2.[5] The cup has been observed by Bednarski[6] to be absent in 95 percent of high myopes and two thirds of hyperopes, but only in 14 percent of emmetropes. However, axial myopia is, in general, associated with a larger C/D ratio than is hyperopia.[7] For reference, the incidence of refractive errors in an unselected general ophthalmic population[8] is shown in Table 1-1.

Cup size has been observed by some to increase slightly with age,[1,9] but this is disputed by others.[5,8] Increased intraocular pressure and a decreased aqueous outflow (C value) appear to be associated with larger cup/disc ratios in normal

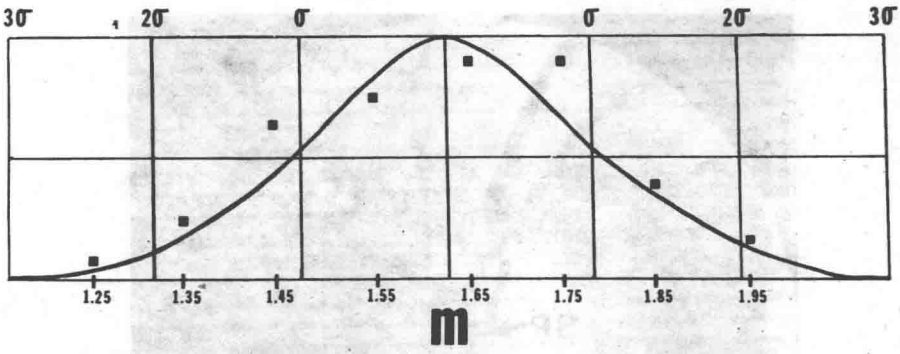

Fig. 1-2. Frequency of distribution of optic nerve head diameters as calculated from averaging horizontal and vertical measurements. The mean of 1.62 mm is designated by the letter m and the diameters are shown in millimeters on the x axis. The small squares represent values found in a group of 100 normal eyes, as compared with the line, which indicates the ideal distribution curve. (Adapted from Franceschetti A, Bock RH: Megalopapilla: A new congenital anomaly. Am J Ophthalmol 33:227–235, 1950.)

eyes.[10] In contrast to cups of glaucomatous eyes, in which vertical enlargement and extension to the margin of the disc may be present, physiologic cupping is usually round or slightly oval horizontally and is associated with a rim of normal nerve tissue.[11]

The central retinal artery enters the globe through the optic nerve head, commonly nasal to the site of exit of the central retinal vein (Figs. 1-1 and 1-4). On the papilla the artery most often divides into superior and inferior papillary

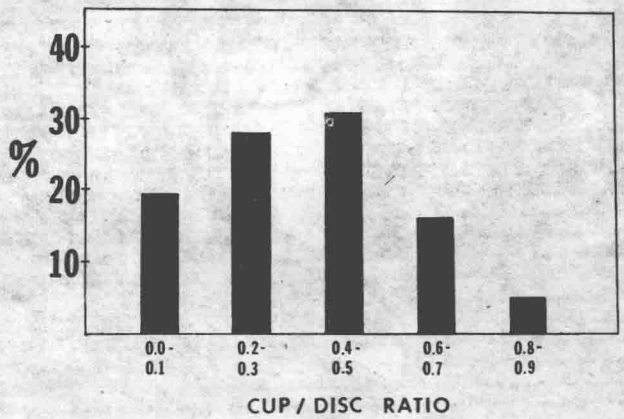

Fig. 1-3. Distribution of cup/disc ratios in normal eyes. (Data from Armaly MF: The optic cup in the normal eye. Am J Ophthalmol 68:401–407, 1969.)

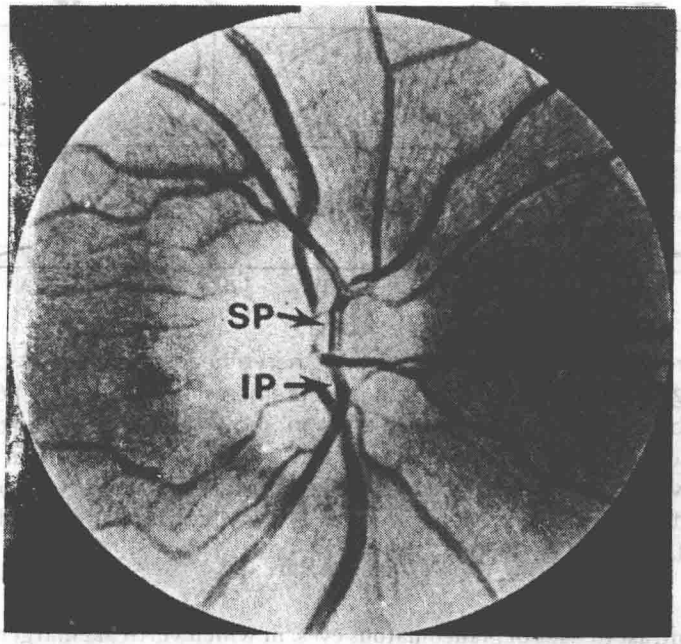

Fig. 1-4. Optic disc showing emergence of the retinal arteries nasal to their venous counterparts. The central retinal artery divides into a superior papillary branch (SP) and an inferior papillary branch (IP), which in turn split into quadrantic branches.

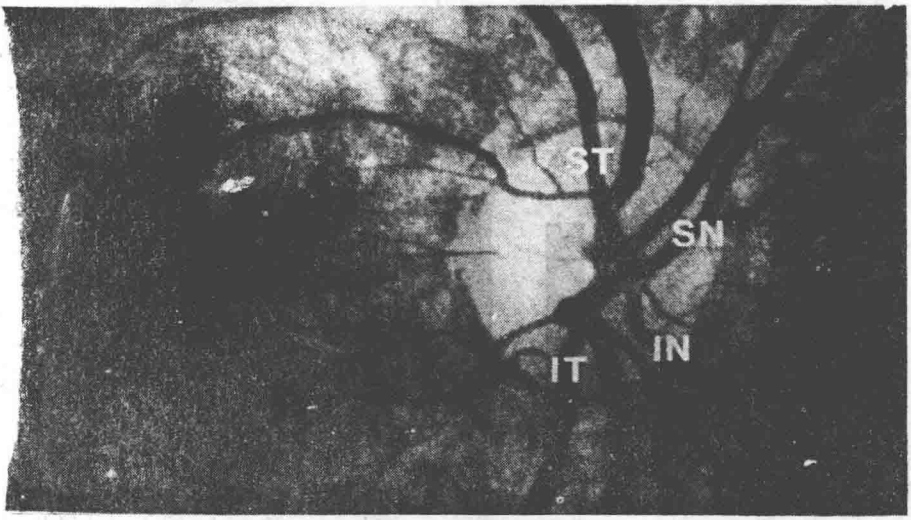

Fig. 1-5. Superotemporal (ST), superonasal (SN), inferotemporal (IT), and inferonasal (IN) branch retinal arteries entering at the same point on the optic disc.

branches (Fig. 1-4), which in turn divide to supply the four quadrants of the fundus. Sometimes the four quadrantic branches enter together on the disc (Fig. 1-5). Occasionally, the central artery divides within the substance of the optic nerve itself, so that the branches can then be seen emanating separately on the disc. The central retinal vein is analogous in that it is usually formed by an inferior and a superior branch, each derived from two quadrantic branches. A common variant seen on the nerve head is the presence of a cilioretinal artery emerging to supply a portion of the retina. As this has been described with fluorescein angiography in up to 32 percent of eyes,[12] it can almost be considered a variant of normal, rather than abnormal.

In many eyes the optic disc directly adjoins the orange-appearing fundus along its entire border. However, in about 25 percent of eyes a juxtapapillary crescent is present.[13] While the crescent may occur anywhere along the margin of the disc, 87 percent are located temporally (Fig. 1-6). About two thirds of the remainder are located inferiorly. Most temporal crescents are myopic and are acquired during life due to a continous growth process. True congenital crescents are present at birth and remain unchanged throughout life. The incidence of different crescents is

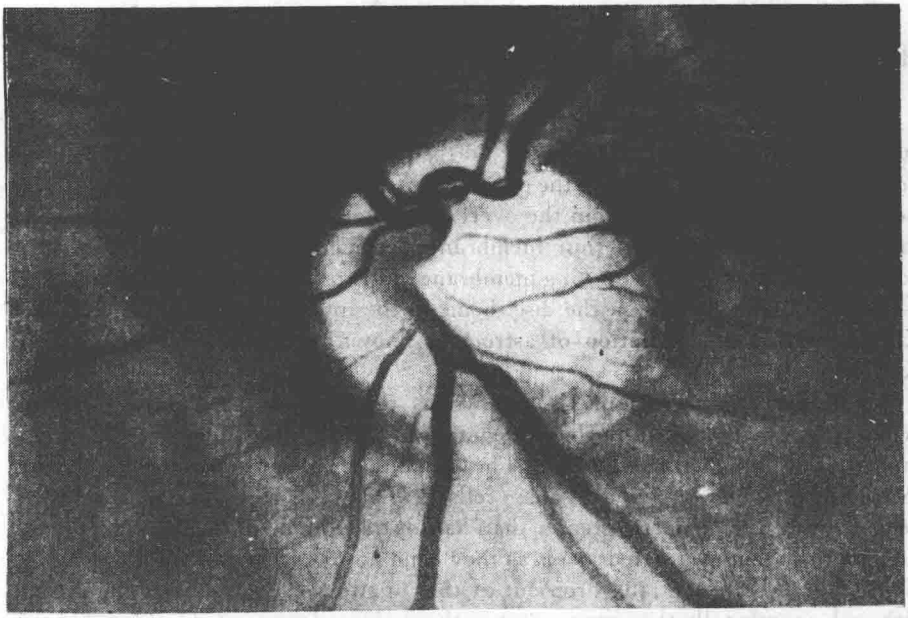

Fig. 1-6. Temporally located juxtapapillary crescent, characterized by a relative absence of choroid and retinal pigment epithelium over the defect. Most crescents of this variety occur in myopic eyes and are acquired.

Table 1-2
Incidence of Juxtapapillary Crescents in All Eyes

Location	Percentage
Temporal	87
Inferior	9
Superior	2
Nasal	2

Data from Mann.[13]
The majority of temporal crescents are acquired and present in myopic eyes, while those located elsewhere are most commonly congenital.

shown in Table 1-2. Histologically, crescents are characterized by variable degrees of absence of the juxtapapillary choroid, retinal pigment epithelium (RPE), and outer retina. Occasionally, hyperplasia of the RPE can also be seen.

HISTOLOGY

The optic nerve head can be conveniently divided into three portions, the surface layer, the prelaminar region, and the lamina cribrosa.[14] The surface layer consists of nerve fibers entering the optic nerve from all regions of the retina. These nerve fibers are separated from the overlying vitreous body by the inner limiting membrane of Elschnig, a thin membrane derived from astrocytes, which is continuous with the inner limiting membrane of the retina at the disc margin (Fig. 1-7).[15] Located centrally on the disc, beneath the internal limiting membrane of Elschnig, is an accumulation of astrocytes known as the central meniscus of Kuhnt.

The prelaminar region, also known as the anterior or choroidal lamina cribrosa, is characterized by the segregation of retinal nerve fibers into approximately 1000 bundles (fascicles).[16] The bundles are separated by a framework formed by astroglia known as "spider cells."[17] This supporting glial structure has been estimated to comprise greater than half of the volume of the papilla. It lends support to the unmyelinated axons as they bend at 90° angles at the disc margin. These astrocytes essentially drop out at the junction of the disc and the retina, although occasionally they are present in the peripapillary nerve fiber layer of the retina. Encircling the prelaminar region of the optic nerve, but much more developed on the temporal side, is the border tissue of Elschnig, which is

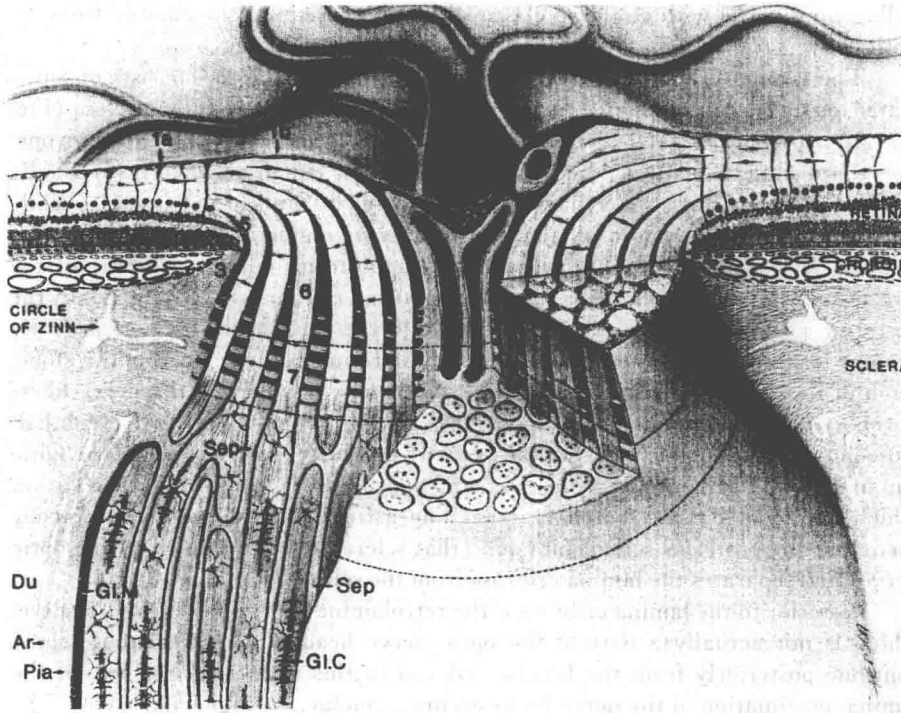

Fig. 1-7. Three-dimensional schematic of the anterior portion of the optic nerve. The numbered regions include (1a) the inner limiting membrane of the retina, which is continuous with (1b) the inner limiting membrane of Elschnig; (2) the central meniscus of Kuhnt, an area of astrocytic and connective tissue thickening centrally beneath the inner limiting membrane of Elschnig; (3) the border tissue of Elschnig, a rim of collagenous connective tissue that extends anteriorly from the pia-scleral junction to separate choroid and optic nerve tissue; (4) the border tissue of Jacoby, which is astroglial tissue lining the scleral canal through which the optic nerve enters the globe; (5) the intermediary tissue of Kuhnt (anterior extension of the glial tissue of Jacoby), separating the outer retinal layers from the optic nerve; (6) the prelaminar region (anterior lamina cribrosa); (7) the lamina cribrosa (posterior lamina cribrosa). The dura (Du), arachnoid (Ar) and pia mater (Pia) are shown, as is the glial mantle (Gl.M) of astrocytes, which surrounds the nerve and is continuous with the border tissue of Jacoby. Connective tissue septae (Sep) separate the nerve fiber bundles. Within the retrolaminar nerve bundles, astrocytes and oligodendrocytes form columns of nuclei (Gl.C). (From Anderson DR, Hoyt WF: Ultrastructure of intraorbital portion of human and monkey optic nerve. Arch Ophthalmol 82:506–530, 1969. With permission. Photograph courtesy of Dr. Douglas Anderson.)

collagenous tissue with glial and elastic elements separating the choroid from the optic nerve.

The lamina cribrosa region, or posterior lamina cribrosa, consists of fenestrated, or sievelike, connective tissue continuous with the surrounding sclera (Fig. 1-7). In hyperopic eyes it lies 0.7 mm behind the surface of the retina, and in myopic eyes this distance is halved.[16] Because of this, the disc cup in myopic eyes is usually not deep, thereby making the diagnosis of glaucoma difficult in some cases. (While certain authors have chosen to label the prelaminar region as the anterior lamina cribrosa,[15,17,18] and the sievelike network as the posterior lamina cribrosa, for the purpose of clarity in this book the term *lamina cribrosa* will refer to only the posterior portion.) The collagenous connective tissue in this region provides support for the axons of the ganglion cells of the retina as they exit from the globe. Each of the fenestrae has an astroglial lining, and consequently the nerve fibers normally do not directly come into contact with the connective tissue.[19] Ophthalmoscopically, the supportive sievelike network can be seen at the base of the optic cup in about 34 percent of normal eyes[4] and appears as yellow-white tissue within which are located many small craters. The astroglial border tissue of Jacoby surrounds the posterior scleral foramen (that scleral defect occupied by the optic nerve) and separates the lamina cribrosa from the sclera.

Posterior to the lamina cribrosa is the retrolaminar portion of the optic nerve, which is not actually a part of the optic nerve head. The collagenous septae continue posteriorly from the lamina cribrosa in this area, and just behind the lamina, myelination of the nerve fibers occurs secondary to oligodendrocytes. The vaginal sheaths are also acquired at this point, and the nerve thickens substantially.[20]

VASCULAR SUPPLY

The blood supply to the optic nerve head is derived from both the central retinal and posterior ciliary arterial circulations. The surface nerve fiber layer of the nerve is supplied essentially by retinal arterioles that arise on the disc and from recurrent branches that originate in the circumpapillary region.[14] When a cilioretinal artery is present, however, the superficial capillaries may arise from this vessel on the sector of the disc in which it is located. The surface capillaries may be continuous with those in peripapillary retina.[14] Controversy exists as to whether radial peripapillary capillaries (those superficial retinal capillaries which extend outward from the optic disc in a double Bjerrum arcuate fashion) arise from surface vessels on the disc[21] or from peripapillary retinal arterioles.[22]

The blood supply to the prelaminar region is probably derived mainly from centripetal branches of choroidal vessels surrounding the nerve head (Fig. 1-8).[14,18,23,24] While some evidence suggests that it may originate directly from the short posterior ciliary arteries,[25] most authors[14,18,23,24] agree that the major contribution comes directly from the choroid.