CHAPTER 11

Retina

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In this chapter we discuss diseases primarily affecting the sensory retina and retinal pigment epithelium. Many of the degenerative and dystrophic processes described also affect adjacent structures, e.g., the uvea. Moreover, many inflammatory processes which primarily affect the uvea may involve the sensory retina as well. These have been described in Chapter 4.

Basic information and reading include: Balbantyne and Michaelson,55 Blodi et al.,78 Eisner,197 Michaelson,536 Sautter et al.,691 Walsh and Hoyt,528 Wessing,837 Wise et al.858

Congenital Anomalies and Hereditary Diseases

Hypoplasia and Dysplasia of the Retina

Hypoplasia

Hypoplasia is defined as a decrease in the number of retinal ganglion cells and axons. This very rare condition is usually associated with gross anomalies of the central nervous system, e.g., anencephaly (see Chapter 13).

Dysplasia

The terms “dysplasia” and “dystrophy” are strictly defined and denote various processes which are often difficult to distinguish. “Dysplasia” signifies a maldevelopment of embryonic tissues which are pluripotential, creating a bizarre type of growth. Retinal dystrophy signifies a regression or atrophy (abiotrophy) of previously formed tissues, usually caused by hereditary influences. A degeneration typically represents a breakdown of tissues due to exogenous influences, e.g., aging, inflammatory processes, and vascular disease.

Exposure to any noxious agent, e.g., toxic pharmacologic agents or x-rays, during the early months of pregnancy leads to a disturbance of embryogenesis. This creates a more or less chaotic orientation and distribution of retinal cells, leading to the formation of characteristic dysplastic retinal rosettes. These represent an abortive attempt to form embryonic retinal photoreceptors. The rosette consists of rudimentary rods and cones surrounding a central space lined by a membranelike structure analogous to the external limiting membrane of the sensory retina. In addition to rosette formation, retinal dysplasia may also be characterized by proliferation of nonspecific glial and undifferentiated ciliary epithelium. The differential diagnosis for retinal dysplasia is listed in Table 11-1.

Many authors designate retinal “dysplasia” as the retinal changes and folds seen in such conditions as traction retinal detachment, congenital or infantile inflammations, or trauma. It is preferable to confine the term dysplasia to true maldevelopment of the retina with formation of complete, well-formed rosettes.

The pathogenesis and structure of Flexner-Wintersteiner rosettes are similar to those of congenital dysplastic rosettes (see Fig. 11–34). The rosettes in retinoblastomas, of course, have features of atypism and anaplasia (indicative of malignancy) which are absent in dysplastic rosettes.

For further information see: Albert et al.,16 Davson,168 Eagle et al.,192 Fulton et al.,269 Hunter and Zimmerman,382 LaHav et al.,466
Table 11–1. Differential Diagnosis of "Retinal Dysplasia"

<table>
<thead>
<tr>
<th>Cause</th>
<th>Description</th>
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<tr>
<td>Trisomy 13</td>
<td>Associated with colobomas of the fundus and within colobomatous cysts</td>
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<tr>
<td>Norrie’s disease</td>
<td>Associated with falciform retinal fold</td>
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<tr>
<td>Associated with various types of microphthalmia</td>
<td>After irradiation to the embryo, after trauma, hemorrhage, or inflammation during the early months of pregnancy</td>
</tr>
<tr>
<td>Idiopathic, isolated, and unilateral</td>
<td>Reese and Blodi, Reese and Straatsma, and Silverstein et al.</td>
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Myelinated (Medullated) Retinal Nerve Fibers

Myelination of ganglion cell axons begins during fetal life and progresses from the lateral geniculate body toward the globe. The optic tracts are therefore the first portions to be ensheathed by myelin. The fibers of the chiasm and the optic nerve itself are the last portions to be myelinated. When myelination is completed shortly after birth, the myelin sheaths extend up to the posterior aspect of the lamina cribrosa. The axons anterior to the lamina cribrosa, as well as of the nerve fiber layer of the retina, are normally unmyelinated. Occasionally, as a developmental variation, myelination of the nerve fibers extends into the nerve fiber layer of the peripapillary retina. This is usually of no clinical significance. A small scotoma may subtend the affected area and is due to opacification of the superficial retina by the myelinated fibers. The white material retards entrance of light into the retina.

The area of medullated nerve fiber layers may be small and isolated or occasionally affect multiple areas of the fundus or the entire surface of the optic nerve (Fig. 11–1). The myelin substance overlies and therefore obscures the retinal vessels. When large areas of the fundus are myelinated, a right pupillary reflex may occur, creating a clinical leucocoria.

The differential diagnosis of myelinated nerve fibers includes the various forms of leukocoria, intraocular tumors (including retinoblastoma and various phakomatoses), and remnants of Bergmeister’s papilla (see Chapter 10). Usually medullated nerve fibers remain unchanged throughout life. However, any disease...