CHAPTER 1

Introduction to Part I

This chapter includes related activities.

Indicates that supplemental figures are available in the Pathology Atlas

Highlights

- Ophthalmic pathology is a subspecialty recognized by the American Academy of Ophthalmology, as well as other North American and international ophthalmology organizations.
- Pathologic entities are generally divided into the following major categories: developmental anomaly, inflammation, dystrophy and degeneration, and neoplasia.
- Developmental anomalies usually involve abnormalities in size, location, organization, or amount of tissue.
- Inflammation can be broadly classified as acute or chronic, focal or diffuse, granulomatous or nongranulomatous, and infectious or noninfectious.
- The degeneration of a blind, traumatized eye involves a range of tissue alterations, including atrophia bulbi without shrinkage, atrophia bulbi with shrinkage, and phthisis bulbi.

Overview

Ophthalmic pathology is recognized as a subspecialty by the American Academy of Ophthalmology, the American Board of Ophthalmology, the Association of University Professors of Ophthalmology, and the International Council of Ophthalmology. The study of ophthalmic pathology has contributed significantly to our understanding of the pathogenesis of diseases of the eye and ocular adnexa. In the United States, ophthalmologists and pathologists may receive subspecialty fellowship training in ophthalmic pathology after completion of an ACGME (Accreditation Council for Graduate Medical Education)—accredited residency in ophthalmology or pathology; some ophthalmic pathologists are board certified in both ophthalmology and pathology.
Clinical Pearl  Ophthalmic pathology plays an important role in the diagnosis of various ophthalmic diseases, in cancer staging, and in guiding clinical management.

BCSC Section 4, *Ophthalmic Pathology and Intraocular Tumors*, provides a general overview of ophthalmic pathology and oncology: common practices and pathologic processes, as well as some less common, but important, entities, are discussed. For more comprehensive reviews of these entities, please refer to the references listed in the Additional Materials and Resources section at the end of this volume.

This chapter describes the organizational framework used in Chapters 4 through 14. Chapter 2, the Quick-Start Guide, discusses specimen handling and processing and emphasizes the importance of communication between the ophthalmologist and the pathologist for providing good patient care. Chapter 2 also covers special testing modalities such as immunohistochemical staining, flow cytometry, and frozen sections. Chapter 3 discusses the basic principles and specific aspects of wound repair. The remainder of Part I, Chapters 4 through 14, is dedicated to specific anatomical regions and pathology.

Clinical Pearl  Communication between the ophthalmologist and pathologist is essential for optimal patient care. In especially complex cases, the ophthalmologist is encouraged to review pathology specimen slides with the pathologist.

Organizational Framework and Basic Pathologic Concepts

As stated previously, Chapters 4 through 14 in this volume focus on specific ocular structures and disease processes. In these chapters, the text is organized from general to specific, with the following topics as the framework for the discussion:

- topography
- disease process
- differential diagnosis

Thus, the text also provides an organizational paradigm for the study of ophthalmic pathology (Table 1-1).

Topography

When used in ophthalmic pathology, the term *topography* refers to the description of the anatomical location and structural features of a particular tissue. Topographic identification is the first step in analysis of a pathologic specimen. Identifying normal tissue in a specimen helps clinicians to define the abnormal areas and narrow the differential diagnosis.
Understanding the disorders that affect the eye and periocular structures requires an understanding of normal ocular structures and functions (Activities 1-1, 1-2).

**ACTIVITY 1-1** Topography of the eye and orbit: identify structures.
*Developed by Tatyana Milman, MD.*
*Available at: aao.org/

**ACTIVITY 1-2** Histology of the eye and orbit: identify tissues.
*Developed by Tatyana Milman, MD.*
*Available at: aao.org/

Using the topographic features of the specimen, an examiner can orient and identify the tissue in question. For example, collagenous tissue lined by keratinized stratified squamous epithelium with dermal appendages is typical of eyelid skin, whereas organized layers, including nonkeratinized stratified squamous epithelium, Bowman layer, collagenous
stroma, Descemet membrane, and endothelium, are typical of the cornea. Recognition of characteristic features, such as the presence or absence of an epithelium, can be particularly helpful. See BCSC Section 2, *Fundamentals and Principles of Ophthalmology*, for a review of ophthalmic anatomy.

### Disease Process

In the evaluation of a pathologic specimen, the examiner should attempt to determine the general disease process after surveying the topography (Activity 1-3). The major disease processes discussed in Chapters 4 through 14 include

- developmental anomaly
- inflammation
- dystrophy and degeneration
- neoplasia

**ACTIVITY 1-3**  Identify the disease process.

*Developed by Vivian Lee, MD.*

Available at: aao.org

#### Developmental anomaly

Developmental anomalies are structural or functional anomalies that develop in utero or during early childhood, while the body is developing. They may be detected prenatally, at birth, or later in life. Developmental anomalies usually involve abnormalities in the size, location, organization, or amount of tissue, such as those seen in congenital hypertrophy of the retinal pigment epithelium or eyelid coloboma. Often, these anomalies are classified as choristomas or hamartomas. A choristoma consists of normal, mature tissue (1 or 2 embryonic germ layers) at an abnormal location. An epibulbar dermoid is classified as a choristoma because it consists of normal, mature skin structures at an atypical location, the limbus. In contrast, the term hamartoma describes hypertrophy and hyperplasia (abnormal amount) of mature tissue in a normal location. One example of a hamartoma is an orbital cavernous venous malformation, which is an encapsulated mass of mature venous channels in the orbit. A tumor made up of tissue derived from all 3 embryonic germ layers is called a teratoma (Fig 1-1).

#### Inflammation

Inflammation can be classified in several ways (see Table 1-1), for example:

- **onset**: acute or chronic
- **location**: focal or diffuse
- **predominant cell type**: granulomatous or nongranulomatous
- **etiology**: infectious or noninfectious

A bacterial corneal ulcer, for instance, is generally an acute, focal, nongranulomatous inflammatory process, whereas sympathetic ophthalmia is a chronic, diffuse, granulomatous inflammatory disease.
In the early phases of the inflammatory process, polymorphonuclear leukocytes, which include neutrophils, eosinophils, and basophils, typically predominate (Fig 1-2). Neutrophils typify the acute inflammatory response and can be recognized by their multi-segmented nuclei and intracytoplasmic granules (Fig 1-3). They are often associated with bacterial infections and may be found in blood vessel walls in some forms of vasculitis. Eosinophils are commonly associated with allergic reactions but may also be present in chronic inflammatory processes such as sympathetic ophthalmia. They have bilobed nuclei and prominent intracytoplasmic eosinophilic granules (Fig 1-4). Basophils and mast cells are also involved in allergic responses and contain basophilic intracytoplasmic granules (Fig 1-5). Mast cells are difficult to identify in tissue sections without the assistance of special stains.