

CONTENTS

Preface	ix
Theresa A. Beery	

Genetics in Critical Care: A Toolbox	139
Theresa A. Beery	

Becoming comfortable with the onslaught of genetic information requires a familiarity with genetics terminology and basic molecular genetics. Caring for the genetic health of our patients requires an additional appreciation of genetic testing and screening issues and new areas of study with strange-sounding names, such as proteomics, nutrigenomics, and epigenetics. This article provides an overview of basic genetic principles and terminology with applications to critical care patients. It lays a foundation to support understanding of the other articles in this issue. An appreciation of genetics (the study of heredity) and genomics (the study of the interaction of genes within an organism) has an important place in the knowledge base of every critical care clinician.

The Family History: Reemergence of an Established Tool	149
Robert B. Hinton Jr.	

Genetics has transformed the use of family history information and has led to the reemergence of the detailed genetic family history. It is critical that public and professional educational efforts to increase family history awareness and working knowledge are prioritized. Patient maintenance of the pedigree provides increased patient awareness and facilitates some of the limitations associated with conventional medical history ascertainment, ultimately improving health care and research. The increasing use of genetic screening promises to cultivate a paradigm shift in medical treatment emphasizing primary prevention and early intervention. Appreciation of the family history is necessary to make this important advance.

Congenital Heart Disease and Genetics	159
Vicki L. Zeigler	

Improvements in the diagnosis and treatment of congenital heart disease have drastically reduced the morbidity and mortality associated with such defects. Knowledge regarding the genetic contributions to congenital heart disease is considered to be in its infancy; however, the field of cardiovascular genetics in humans is moving at a rapid pace. This article discusses what is currently known about the genetic contribution to congenital heart disease, including structural defects and congenital cardiac arrhythmias. Genetic and chromosomal syndromes that involve the heart are reviewed along with genetic testing.

Genetic Influences in Common Respiratory Disorders M. Linda Workman and Chris Winkelman	171
<p>Respiratory disorders are common problems for adults and children in North America and generally represent the outcome of gene–environment interactions. Some problems are considered genetic in origin, such as cystic fibrosis, and others are considered environmental in origin, such as respiratory infections. Emerging information indicates that even genetic-based disorders are influenced by the environment and that environmental-based disorders are modified by personal genetic factors in individual physiologic responses. An understanding of an individual’s personal risk factors for disease or health problem development can allow health care professionals to tailor health promotion strategies and treatment plans with appropriate environmental manipulation. This article explores the genetic influences that may affect the individual’s physiologic responses and the consequences of environmental stimuli.</p>	
Hereditary Hemochromatosis: Pathophysiology, Diagnosis, and Management Christopher Fowler	191
<p>Hereditary hemochromatosis (HH) is an autosomal recessive genetic disease resulting in inappropriate intestinal iron absorption leading to iron overload and end-organ disease. The disease is most prevalent in white individuals of European descent. The C282Y mutation on the HFE gene accounts for most cases of HH; however, other genetic mutations have been identified. End-organ damage results in cirrhosis, diabetes mellitus, and cardiomyopathy. Therapeutic phlebotomy to deplete excessive iron stores is the standard treatment of HH and results in normal longevity if therapy is initiated before end-organ disease occurs.</p>	
Genes and Acute Neurologic Disease and Injury: A Primer for the Neurologic Intensive Care Nurse Sheila A. Alexander and Michael Beach	203
<p>The genetics revolution has not evaded the neuroscience community. Neurologic diseases and injuries, particularly of the type seen in neurologic intensive care units, are difficult to treat and often have poor prognoses. Recent work has begun to identify genotype-specific influences on development and treatment of multiple sclerosis and stroke. Additionally, responses to diseases and injuries to the brain and spinal cord have genetic influences. This article informs nurses working with neurologically impaired patients in the intensive care unit of specific genes involved in patient response and potential future therapeutics.</p>	
Inflammation and Genomics in the Critical Care Unit Chris Winkelman	213
<p>Inflammation is a physiologic response to irritants, injury, and infection. Inflammatory dysregulation is believed to contribute to mortality and morbidity in illnesses common to critical care units, such as burns, trauma, sepsis, and cardiovascular disease. This article reviews the approaches used to investigate the molecular basis of inflammatory function. Genomic findings are providing insight into clinical diagnosis and treatment of inflammatory derangements in critically ill patients.</p>	
Pharmacogenetics in Critical Care: Atrial Fibrillation as an Exemplar Cynthia A. Prows and Theresa A. Beery	223
<p>Pharmacogenetic testing is currently not routine in critical care settings but recent changes in the warfarin label are likely to lead to critical care nurses encountering physician or nurse practitioner orders for such testing. Although the science for</p>	

pharmacogenetics is complex, the components of patient teaching are not beyond that which nurses already provide about other laboratory, disease, and treatment-based information. It is reasonable to expect that as the science of pharmacogenetics and pharmacogenomics expands and discoveries are translated in clinical settings, the additional information from pharmacogenetic test results will help prescribers select or adjust medication doses to reduce the risk for adverse drug reactions and improve the chances of achieving therapeutic targets in a timely fashion.

Neonatal Genetic Testing is More Than Screening

233

Carole Kenner, Judith A. Lewis, Jana L. Pressler, and Cindy M. Little

Newborn screening practices have changed since breakthroughs have occurred in genetics and mapping of the human genome. Although newborn screening has been in existence since the 1960s, today's newborn screening practices are subsumed primarily under the umbrella of genetic testing. Inclusion of the family history tool is another dimension of neonatal assessment. Technology allows many noninvasive tests to be run at a low cost but with this advance comes ethical and legal dilemmas. This article discusses neonatal genetic testing and some of the ethical dilemmas that arise.

Index

239