

Disorders

Some disease resulting from or attributed to abnormalities of Membranes

Disease	Abnormality
Achondroplasia (MIM 100800)	Mutations in the gene encoding the fibroblast growth factor receptor 3
Familial hypercholesterolemia (MIM 143890)	Mutations in the gene encoding the LDL receptor
Cystic fibrosis (MIM 219700)	Mutations in the gene encoding the CFTR protein, a Cl^- transporter
Congenital long QT syndrome (MIM 192500)	Mutations in genes encoding ion channels in the heart
Wilson disease (MIM 277900)	Mutations in the gene encoding a copper-dependent ATPase
I-cell disease (MIM 252500)	Mutations in the gene encoding GlcNAc phosphotransferase, resulting in absence of the Man 6-P signal for lysosomal localization of certain hydrolases
Hereditary spherocytosis (MIM 182900)	Mutations in the genes encoding spectrin or other structural proteins in the red cell membrane
Metastasis	Abnormalities in the oligosaccharide chains of membrane glycoproteins and glycolipids are thought to be of importance
Paroxysmal nocturnal hemoglobinuria (MIM 311770)	Mutation resulting in deficient attachment of the GPI anchor to certain proteins of the red cell membrane

Red Cell Membrane Transport In Health And Disease

**Johann S. Ach, Beate
Lüttenberg, Rainer Paslack, Klaus-
Michael Weltring**



Red Cell Membrane Transport In Health And Disease:

Red Cell Membrane Transport in Health and Disease Ingolf Bernhardt, J. Clive Ellory, 2013-04-17 The red cell has been a focus for scientific and medical investigation since the earliest times. A higher erythrocyte sedimentation rate was associated with diseases usually pyrexias before the thermometer was invented. Furthermore ever since the early observers Swammerdam and Leeuwenhoek saw discrete corpuscles in samples of blood using the first microscopes there has been a significant scientific interest in the structure and function of red blood cells. The later discovery that red cells were not spherical but biconcave discs introduced a scientific puzzle which is still not completely resolved today and identified the need for a detailed knowledge of the plasma membrane composition and structure and its interaction with the cytoskeleton. Important concepts like the lipid bilayer together with its more recent refinement as asymmetric in phospholipid composition led to the identification of translocases involved in actively maintaining its composition. Understanding the mechanics of red cell deformation as these biconcave discs traverse capillaries was advanced by the pioneering work of Rand and Burton in the Sixties and progressed by Evans, Skalak and others. Based on the bilayer couple hypothesis the shape changes that are possible for a human red cell from echinocyte to stomatocyte were described by Sheetz and Singer in the Seventies in terms of alterations in the individual halves of the bilayer. Certain clinical conditions are associated with obvious changes in red cell morphology. **Erythrocytes: Physiology And Pathophysiology** Florian Lang, Michael Foller, 2012-07-23 The book covers the functional significance and properties of erythrocytes their generation, senescence and suicidal death. It further summarizes knowledge about hormones influencing erythrocyte formation including erythropoietin as well as disorders affecting and involving erythrocytes such as anemia, malaria and sepsis. This seminal work forms a unique reference on the most abundant cell type in mammals and will be an invaluable resource for students in the life sciences. **Research Awards Index**, 1989 **Subject Index of Current Research Grants and Contracts Administered by the National Heart, Lung and Blood Institute** National Heart, Lung, and Blood Institute, 1977 **Calcium signalling** Lars Kaestner, 2012-12-02 The book combines general concepts and methods to investigate calcium signalling in cells ranging from molecular biology approaches to manipulation of calcium in living cells. The focus within these methods is on the broad range of fluorescence imaging technology in particular on optical sectioning techniques and fast image acquisition. In addition to these general guidelines there are application examples in a context beyond calcium signalling in two major fields: investigations of isolated cardiac myocytes and red blood cell related research. While the cellular cardiology section provides snapshots of certain calcium signalling aspects the red blood cell part presents an overview from the functional identification of calcium channels to a concept of physiological and pathophysiological relevance. **Research Grants Index** National Institutes of Health (U.S.). Division of Research Grants, 1975 **The Red Cell Life-Cycle From Erythropoiesis to Clearance** Lars Kaestner, Anna Bogdanova, 2019-02-05 The eBook The red cell life cycle from erythropoiesis to clearance continues the discussion of

questions like What are the changes associated with red blood cell maturation adulthood and senescence What are the determinants of red blood cell life span and clearance What are the mechanisms in control of red blood cell mass in healthy humans and patients with various forms of anaemia Can red blood cells be trained to provide the body with more oxygen during endurance exercises What are the markers of circulating red blood cell senescence and in cells during storage and transfusion And what can be learned from various species that developed advanced adaptations to maintain oxygen delivery under stress conditions such as exercising to the limit diving or living in anaerobic aquatic habitats or at high altitude Within the approximately 120 days or 40 in a mouse or 150 170 in a horse life span of healthy red blood cells many cellular properties change leading to aged mixed cell populations in the circulation Red blood cells seem to be genetically terminated by the time they become red blood cells and the contributions of this eBook increase the understanding of this process There are surprisingly versatile remodeling processes happening during the red blood cell life span Numerous disorders are associated with the premature onset of the ageing process of red blood cells Furthermore in vitro ageing and or modifications as well as the slowing down of the modifications is an important issue in transfusion medicine Many of the molecular mechanisms behind such effects are elucidated in this eBook

Biomedical Index to PHS-supported Research, 1988 *The Future of Physiology: 2020 and Beyond* George E. Billman, Geoffrey A. Head, 2021-07-30 This Research Topic eBook includes articles from Volume I and II of The Future of Physiology 2020 and Beyond series Research Topic The Future of Physiology 2020 and Beyond Volume I Research Topic The Future of Physiology 2020 and Beyond Volume II The term Physiology was introduced in the 16th century by Jean Francois Fernel to describe the study of the normal function of the body as opposed to pathology the study of disease Over the ensuing centuries the concept of physiology has evolved and a central tenet that unites all the various sub disciplines of physiology has emerged the quest to understand how the various components of an organism from the sub cellular and cellular domain to tissue and organ levels work together to maintain a steady state in the face of constantly changing and often hostile environmental conditions It is only by understanding normal bodily function that the disruptions that leads to disease can be identified and corrected to restore the healthy state During the summer of 2009 I was invited by Dr Henry Markram one of the founders of the Frontiers In series of academic journals to serve as the Field Chief Editor and to launch a new Open access physiology journal that would provide a forum for the free exchange of ideas and would also meet the challenge of integrating function from molecules to the intact organism In considering the position I needed to answer two questions 1 What exactly is Open access publishing and 2 What could Frontiers in Physiology add to the already crowded group of physiology related journals As a reminder the traditional model of academic publishing is a process by which academic scholars provide material reviewing and editing expertise for publication free of charge then pay to publish their work and to add insult to injury they and their colleagues must pay the publisher a fee either directly or via an institutional subscription to read their published work

slightly modified from the The Devil's Dictionary of Publishing Physiology News the quarterly newsletter of the Physiological Society Spring 2019 Issue 114 page 8 In the traditional model the publisher not the authors owns the copyright such that the author must seek permission and may even be required to pay a fee to re use their own material such as figures in other scholarly articles reviews book chapters etc In contrast individuals are never charged a fee to read articles published in open access journals Thus scholars and interested laymen can freely access research results that their tax dollars paid for even if their home institution does not have the resources to pay the often exorbitant subscription fees Frontiers takes the open access model one step further by allowing authors rather than the publisher to retain ownership i e the copyright of their intellectual property Having satisfied the first question I then considered whether a new physiology journal was necessary At that point in time there were no open access physiology journals and further many aspects of physiology were not covered in the existing journals Frontiers afforded the unique opportunity to provide a home for more specialized sections under the general field journal Frontiers in Physiology with each section having an independent editor and editorial board I therefore agreed to assume the duties of Field Chief Editor in November 2009 Frontiers in Physiology was launched in early 2010 and the first articles were published in April 2010 Since these initial publications we have published over 10 000 articles and have become the most cited physiology journal Clearly we must be fulfilling a critical need Now that it has been over a decade since Frontiers in Physiology was launched it is time to reflect upon what has been accomplished in the last decade and what questions and issues remain to be addressed Therefore it is the goal of this book to evaluate the progress made during the past decade and to look forward to the next In particular the major issues and expected developments in many of the physiology sub disciplines will be explored in order to inspire and to inform readers and researchers in the field of physiology for the year 2020 and beyond A brief summary of each chapter follows In chapter 1 Billman provides a historical overview of the evolution of the concept of homeostasis Homeostasis has become the central unifying concept of physiology and is defined as a self regulating process by which a living organism can maintain internal stability while adjusting to changing external conditions He emphasizes that homeostasis is not static and unvarying but rather it is a dynamic process that can change internal conditions as required to survive external challenges and can be said to be the very basis of life He further discusses how the concept of homeostasis has important implications with regards to how best to understand physiology in intact organisms the need for more holistic approaches to integrate and to translate this deluge of information obtained in vitro into a coherent understanding of function in vivo In chapter 2 Aldana and Robeva explore the emerging concept of the holobiont the idea that every individual is a complex ecosystem consisting of the host organism and its microbiota They stress the need for multidisciplinary approaches both to investigate the symbiotic interactions between microbes and multicellular organisms and to understand how disruptions in this relationship contributes to disease This concept is amplified in chapter 3 in which Pandol addresses the future of gastrointestinal physiology emphasizing advances

that have been made by understanding the role that the gut microbiome plays in both health and in disease Professor Head in chapter 4 describes areas in the field of integrative physiology that remain to be examined as well as the potential for genetic techniques to reveal physiological processes The significant challenges of developmental physiology are enumerated by Burggren in chapter 5 In particular he analyzes the effects of climate change environmentally induced epigenetic modification on phenotype expression In chapter 6 Ivell and Annad Ivell highlight the major differences between the reproductive system and other organ systems They conclude that the current focus on molecular detail is impeding our understanding of the processes responsible for the function of the reproductive organs echoing and amplifying the concepts raised in chapter 1 In chapter 7 Costa describes the role of both circadian and non circadian biological clocks in health and disease thereby providing additional examples of integrated physiological regulation Coronel in chapter 8 provides a brief history of the development of cardiac electrophysiology and then describes areas that require further investigation and includes tables that list specific questions that remain to be answered In a similar manner Reiser and Janssen chapter 9 summarize some of the advancements made in striated muscle physiology during the last decade and then discuss likely trends for future research to name a few examples the contribution of gender differences in striated muscle function the mechanisms responsible of age related declines in muscle mass and role of exosome released extracellular vesicles in pathophysiology Meininger and Hill describe the recent advances in vascular physiology chapter 10 and highlight approaches that should facilitate our understanding of the vascular processes that maintain health our old friend homeostasis and how disruptions in these regulatory mechanisms lead to disease They also stress the need for investigators to exercise ethical vigilance when they select journals to publish in and meetings to attend They note that the proliferation of profit driven journals of dubious quality threatens the integrity of not only physiology but science in general The pathophysiological consequences of diabetes mellitus are discussed in chapters 11 and 12 In chapter 11 Ecelbarger addresses the problem of diabetic nephropathy and indicates several areas that require additional research In chapter 12 Sharma evaluates the role of oxidative damage in diabetic retinopathy and then proposes that the interleukin 6 transsignaling pathway is a promising therapeutic target for the prevention of blindness in diabetic patients Bernardi in chapter 13 after briefly reviewing the considerable progress that has been achieved in understanding mitochondrial function lists the many questions that remain to be answered In particular he notes several areas for future investigation including but not limited to a more complete understanding of inner membrane permeability changes the physiology of various cation channels and the role of mitochondrial DNA in disease In chapter 14 using Douglas Adams' The Hitchhikers Guide to the Universe as a model Bogdanova and Kaestner address the question why a young person should study red blood cell physiology and provide advice for early career scientists as they establish independent laboratories They then describe a few areas that merit further attention not only related to red blood cell function but also to understanding the basis for blood related disease and the

ways to increase blood supplies that are not dependent on blood donors Finally the last two chapters specifically focus on non mammalian physiology In chapter 15 Scanes asks the question are birds simply feathered mammals and then reviews several of the significant differences between birds and mammals placing particular emphasis on differences in gastrointestinal immune and female reproductive systems In the final chapter chapter 16 Anton and co workers stress that since some 95% of living animals species are invertebrates invertebrate physiology can provide insights into the basic principles of animal physiology as well as how bodily function adapts to environmental changes The future of Physiology is bright there are many important and interesting unanswered questions that will require further investigation All that is lacking is sufficient funding and a cadre of young scientists trained to integrate function from molecules to the intact organism George E Billman Ph D FAHA FHRS FTPS Department of Physiology and Cell Biology The Ohio State University Columbus OH United States

Mechanisms and Significance of Cell Volume Regulation Florian Lang, 2006 It is essential for the survival of cells to maintain their volume within certain limits Profound alterations interfere with the integrity of the cell membrane and the cytoskeletal architecture Cells use various methods to adjust osmotic pressure gradients which in turn govern water movement across cell membranes The most rapid and efficient means is ion transport across the cell membrane However to avoid excessive alterations of intracellular ion concentration cells additionally employ organic osmolytes for osmoregulation and modify their metabolism thus generating or disposing osmotically active organic substances But a given cell uses only part of the cell volume regulatory mechanisms at their disposal The large repertoire available allows for a selection of those which have the least untoward impact on cell function The present book is a collection of reviews on the various aspects of cell volume regulation written by internationally leading experts and constitutes a valuable addition to the library of those working in the field

Physiology and Pathology of Chloride Transporters and Channels in the Nervous System F. Javier Alvarez-Leefmans, Eric Delpire, 2009-08-22 The importance of chloride ions in cell physiology has not been fully recognized until recently in spite of the fact that chloride Cl together with bicarbonate is the most abundant free anion in animal cells and performs or determines fundamental biological functions in all tissues For many years it was thought that Cl was distributed in thermodynamic equilibrium across the plasma membrane of most cells Research carried out during the last couple of decades has led to a dramatic change in this simplistic view We now know that most animal cells neurons included exhibit a non equilibrium distribution of Cl across their plasma membranes Over the last 10 to 15 years with the growth of molecular biology and the advent of new optical methods an enormous amount of exciting new information has become available on the molecular structure and function of Cl channels and carriers In nerve cells Cl channels and carriers play key functional roles in GABA and glycine mediated synaptic inhibition neuronal growth and development extracellular potassium scavenging sensory transduction neurotransmitter uptake and cell volume control Disruption of Cl homeostasis in neurons underlies pathological conditions such as epilepsy deafness imbalance brain edema and ischemia pain and neurogenic

inflammation This book is about how chloride ions are regulated and how they cross the plasma membrane of neurons It spans from molecular structure and function of carriers and channels involved in Cl transport to their role in various diseases The first comprehensive book on the structure molecular biology cell physiology and role in diseases of chloride transporters channels in the nervous system in almost 20 years Chloride is the most abundant free anion in animal cells THIS book summarizes and integrates for the first time the important research of the past two decades that has shown that Cl channels and carriers play key functional roles in GABA and glycine mediated synaptic inhibition neuronal growth and development extracellular potassium scavenging sensory transduction neurotransmitter uptake and cell volume control The first book that systematically discusses the result of disruption of Cl homeostasis in neurons which underlies pathological conditions such as epilepsy deafness imbalance brain edema and ischemia pain and neurogenic inflammation Spanning topics from molecular structure and function of carriers and channels involved in Cl transport to their role in various diseases Involves all of the leading researchers in the field Includes an extensive introductory section that covers basic thermodynamic and kinetics aspects of Cl transport as well as current methods for studying Cl regulation spanning from fluorescent dyes in single cells to knock out models to make the book available for a growing population of graduate students and postdocs entering the field

Proceed with Caution? Johann S. Ach, Beate Lüttenberg, Rainer Paslack, Klaus-Michael Weltring, 2012

Nanobiotechnology is still a developing field The results and promises of this technology are not only of scientific and economic importance they also raise grave ethical legal and social questions In this context the so called Precautionary Principle or Vorsorgeprinzip is of high relevance What does it mean to proceed with caution in the field of nanobiotechnology How can the principle be applied and specified Is it a suitable tool for the protection against potentially dangerous effects on the environment and human health What is the status of the Precautionary Principle in international agreements and national legislation Proceed with Caution examines the questions that surround the Precautionary Principle in nanobiotechnology Series Munster Studies on Bioethics Munsteraner Bioethik Studien Vol 12 Seldin and Giebisch's The Kidney Robert J. Alpern, Michael J. Caplan, Orson W. Moe, 2012-12-31 A classic nephrology reference for over 25 years Seldin and Giebisch's The Kidney is the acknowledged authority on renal physiology and pathophysiology In this 5th edition such new and powerful disciplines as genetics and cell biology have been deployed to deepen and widen further the explanatory framework Not only have previous chapters been extensively updated but new chapters have been added to incorporate additional disciplines Individual chapters for example now provide detailed treatment of the significance of cilia the role of stem cells is now given special consideration Finally there has been a significant expansion of the section of pathophysiology incorporating the newer findings of cell biology and genetics If you research the development of normal renal function or the mechanisms underlying renal disease Seldin and Giebisch's The Kidney is your number one source for information Offers the most comprehensive coverage on the market of fluid and electrolyte regulation and dysregulation in 85 completely revised

chapters and 10 new chapters Includes 4 sections 62 chapters devoted to regulation and disorders of acid base homeostasis and epithelial and nonepithelial transport regulation Includes foreword by Donald Seldin and Gerhard Giebisch world renowned names in nephrology and editors of the previous three editions *Advances in Planar Lipid Bilayers and Liposomes* A. Leitmannova Liu, Aleš Iglič, 2009-10-09 *Advances in Planar Lipid Bilayers and Liposomes* Volume 10 continues to include invited chapters on a broad range of topics covering the main arrangements of the reconstituted system namely planar lipid bilayers and spherical liposomes The invited authors present the latest results in this exciting multidisciplinary field of their own research group Many of the contributors working in both fields over many decades were in close collaboration with the late Prof H Ti Tien the founding editor of this book series There are also chapters written by some of the younger generation of scientists included in this series This volume keeps in mind the broader goal with both systems planar lipid bilayers and spherical liposomes which is the further development of this interdisciplinary field worldwide Incorporates contributions from newcomers and established and experienced researchers Explores the planar lipid bilayer systems and spherical liposomes from both theoretical and experimental perspectives Serves as an indispensable source of information for new scientists *Pumps, Channels and Transporters* Ronald J. Clarke, Mohammed A. A. Khalid, 2015-10-12 Describes experimental methods for investigating the function of pumps channels and transporters Covers new emerging analytical methods used to study ion transport membrane proteins such as single molecule spectroscopy Details a wide range of electrophysiological techniques and spectroscopic methods used to analyze the function of ion channels ion pumps and transporters Covers state of the art analytical methods to study ion pumps channels and transporters and where analytical chemistry can make further contributions **The Journal of Experimental Biology** , 2009-06 *Veterinary Clinical Pathology* , 2004 Biomedical Index to PHS-supported Research: pt. A. Subject access A-H , 1994 *Normal and Abnormal Red Cell Membranes* ICN Pharmaceuticals, Inc, 1979 Canadian Journal of Physiology and Pharmacology , 1972

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