Molecular Pathogenesis of MODYs

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Molecular Pathogenesis of MODYs F. M. Matschinsky, M. A. Magnuson, Massimo Porta, 2000-03-21 A timely collection of research papers by 18 leading scientists in the area of biochemical genetics of diabetes mellitus these contributions focus on a well defined class of monogenic forms of the disease i e MODYs 1 5 all caused by specific mutations or deletion of genes that control pancreatic beta cell functions HNF 4alpha 1 glucokinase 2 HNF 1alpha 3 IDX 1 4 and HNF 1beta 5 This volume offers a historical perspective of MODYs and it also contains contributions which deal with the fundamental questions of pancreas and liver development critical for the understanding of the syndrome complex discussed Attempts have been made to highlight results and concepts that may be relevant for understanding type II diabetes in general exemplified by contributions that connect the field of pancreatic islet cell biology and pathology with the newest findings in the area of insulin action e g the role of the insulin receptor and insulin receptor substrates IRS 1 3 Covering a broad and novel approach on the subject of MODYs this book looks into the future of molecular genetic and biochemical research in this area It is recommended reading for basic research scientists in the area of diabetes mellitus academically oriented endocrinologists and clinical researchers Molecular Pathology of Diabetes Mellitus R. D. G. Leslie, 1997 Diabetes already a prevalent disease worldwide is increasing at a rate consistent with a global epidemic The number of people affected is expected to double in the next 15 years to about 200 million The disease is already one of the commonest causes of kidney failure blindness and amputations There is an urgent need to prevent either the development of these complications or the disease itself Dramatic advances in our understanding of the pathogenesis of diabetes have led to the hope that diabetes can indeed be prevented Several major international collaborative projects have been initiated with this aim In this book an outstanding group of international scientists provide a broad outline of the advances in basic science which underpin these projects The book offers a clear comprehensive and up to date account for the interested clinician and scientist and will be invaluable as a reference book for those working in the field *Brook's Clinical Pediatric Endocrinology* Charles G. D. Brook, Peter Clayton, Rosalind Brown, 2011-09-22 A benchmark reference textbook An exceptional editorial team and internationally renowned contributors come together to bring you Brook's Clinical Pediatric Endocrinology This new book is full of practical advice and is essential reading for everyone involved in the care of children and adolescents with endocrine disease and disorders. This outstanding reference book has been fully updated to feature new concepts new investigations and new molecularmechanisms and is full of practical clinical advice Theperfect text for pediatric endocrinologists endocrinologists and pediatricians Endocrinology Adult and Pediatric: Diabetes Mellitus and Obesity E-Book Gordon C Weir, J. Larry Jameson, Leslie J. De Groot, 2013-07-15 Meet the growing challenges of diabetes and obesity management with Endocrinology Adult and Pediatric Diabetes Mellitus and Obesity a new diabetes and obesity eBook from the same expert endocrinologists responsible for the highly acclaimed two volume Endocrinology clinical reference With all of the latest

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Principles of Developmental Genetics Sally A. Moody, 2007-07-19 Unlike anything currently available in the market Dr Sally A Moody and a team of world renowned experts provide a groundbreaking view of developmental genetics that will influence scientific approaches in embryology comparative biology as well as the newly emerging fields of stem cell biology and regenerative medicine Principles of Developmental Genetics highlights the intersection of developmental biology with new revolutionary genomic technologies and details how these advances have accelerated our understanding of the molecular genetic processes that regulates development This definitive resource provides researchers with the opportunity to gain important insights into the clinical applicability of emerging new technologies and animal model data This book is a must have for all researchers in genetics developmental biology regenerative medicine and stem cell biology Includes new research not previously published in any other book on the molecular genetic processes that regulates development Chapters present a broad understanding on the application of animal model systems allowing researchers to better treat clinical disorders and comprehend human development Relates the application of new technologies to the manipulation of stem cells causes ofhuman birth defects and several human disease conditions Each chapter includes a bulleted summary highlighting clinical aspects of animal models Endocrine Diseases of Newborn: Epidemiology, Pathogenesis, Therapeutic Option and Outcome Amanda Lesley Ogilvy-Stuart, Paula Caroline Midgley, 2021-11-10 Textbook of Diabetes Richard I. G. Holt, Allan Flyvbjerg, 2024-02-12 Textbook of Diabetes Classic textbook providing diabetologists and endocrinologists with illustrated and clinically focused content on diabetes Now in its sixth edition the Textbook of Diabetes has established itself as the modern international guide to diabetes Sensibly organized and easy to navigate with exceptional illustrations the textbook hosts an unrivalled blend of clinical and scientific content Written by highly experienced editors and international contributors all of whom have provided insight on new developments in diabetes care These include the most recent guidelines from the European Association for the Study of Diabetes EASD the American Diabetes Association ADA Diabetes

UK and the National Institute for Health and Care Excellence NICE and information on the latest treatment modalities used around the world The textbook includes free access to the Wiley Digital Edition which provides easy to use searching across the book the full reference list with web links illustrations and photographs and post publication updates Sample topics covered in Textbook of Diabetes include Diabetes in its historical and social context covering the history of diabetes past classification and diagnosis of diabetes and the global burden of diabetes Normal physiology covering glucose homeostasis islet function and insulin secretion and glucagon in islet and metabolic regulation Pathogenesis of diabetes covering genetics of diabetes and obesity autoimmune type 1 diabetes and other disorders with type 1 diabetes phenotype Other types of diabetes covering endocrine disorders that cause diabetes pancreatic diseases and diabetes and drug induced diabetes Beautifully illustrated with a clinical focus Textbook of Diabetes provides endocrinologists and diabetologists both consultants specialists and those in training with a fresh and comprehensive clinical resource to consult time and time again The text is also of value to specialist diabetes nurses and researchers in the field Genetic variants and metabolic diseases, volume II Tarunveer Singh Ahluwalia, Kavita Jadhav, Marta Szydlowska, Sarina Kajani, 2024-02-12 Metabolic syndrome MetS is a set of comorbidities that collectively increase an individual s risk of developing cardiovascular disease stroke and type 2 diabetes mellitus T2D Per the World Health Organization WHO MetS is typically characterized by obesity insulin resistance hypertension and hyperlipidemia Building on this point some of the major risk factors for development of MetS include increased weight or an obese phenotype lack of physical activity and genetics Interestingly the last decade has witnessed a deluge of Genome Wide Association Studies GWAS that have linked hundreds of genomic with both collective MetS traits as well as individual metabolic disorders sitting within it Currently in the post GWAS era the focus has shifted to characterization of these novel genomic to establish causality and disease relevance This is being pursued by way of functional validation such as gain and loss of function studies investigating resulting metabolic phenotypes mechanisms and pathways underlying these phenotypes their prevalence and potential for risk stratification across populations and finally identification of therapeutic targets for pharmacological intervention Proceedings of the National Academy of Sciences of the United States of America, 2001 Diabetes Associated with Single Gene Defects and Chromosomal **Abnormalities** F. Barbetti, L. Ghizzoni, F. Guaraldi, 2017-03-21 This volume written by renowned experts provides complete coverage of the main genetic conditions associated with diabetes Divided into five sections it offers insights into genetic defects involving the pancreatic beta cell extreme insulin resistance ciliopathies obesity and glucose metabolism chromosomal defects and other genetic conditions associated with increased susceptibility to diabetes Other topics include the various subtypes of monogenic diabetes such as the neonatal form and the Wolfram syndrome as well as chromosomal defects leading to complex conditions affiliated with diabetes like Trisomy 21 or Prader Willi syndrome There are also chapters dedicated to the poorly explored relationships between metabolism and neurodegenerative disorders like Friedreich

s ataxia and muscular dystrophy This book is a reference for every pediatric and adult endocrinologist and diabetologist even experienced ones with an interest in the intricacies and protean aspects of disorders of glucose metabolism secondary to genetic diseases **Molecular Diagnostics** William B. Coleman, Gregory J. Tsongalis, 2007-10-28 Accompanying CD ROM contains a companion eBook version of Molecular diagnostics for the clinical laboratorian Second edition for downloading and use in the reader s PC or PDA Page 4 of cover **The British National Bibliography** Arthur James Wells, 2000

Diagnostic Pathology: Cytopathology - E-Book Dina R. Mody, Michael J. Thrall, Savitri Krishnamurthy, 2022-11-30 This expert volume in the Diagnostic Pathology series is an excellent point of care resource for practitioners and trainees at all levels of experience and training Covering all aspects of cytology including gynecologic nongynecologic exfoliative fine needle aspiration and imaging it incorporates the most recent scientific and technical knowledge in the field to provide a comprehensive overview of all key issues relevant to today s practice Richly illustrated and easy to use the third edition of Diagnostic Pathology Cytopathology is a visually stunning one stop resource for every practicing pathologist resident student or fellow as an ideal day to day reference or as a reliable training resource Covers all areas of cytopathology including clinical radiologic and immunohistochemical as well as cytopathologic features and molecular correlates where applicable Contains new chapters on ancillary molecular tests specific to thyroid prognostic therapy related immunomarkers in cell blocks and small biopsies Provides new immunohistochemical and molecular coverage including new immunostains and genomic targets Incorporates new reporting terminology such as serous fluid and effusions and updates to existing reporting terminologies Reflects the expanded use of fine needle aspiration for small biopsies FNA B with many more images added and updated throughout Keeps you up to date with current and emerging reporting systems on pancreaticobiliary salivary breast and soft tissue cytology Features more than 3 000 print and online images including carefully annotated histology and gross pathology photos full color illustrations clinical photographs and radiologic images to help practicing and in training pathologists reach a confident diagnosis Includes new videos on such topics as the cytoprepratory process cell transfer cell block and smears collodion bag cell block and more Employs consistently templated chapters bulleted content key facts a variety of tables annotated images pertinent references and an extensive index for quick expert cytopathology reference at the point of care Williams Textbook of Endocrinology E-Book Shlomo Melmed, Richard J. Auchus, Allison B. Goldfine, Clifford J. Rosen, Peter A. Kopp, 2024-04-03 Selected for 2025 Doody's Core Titles with Essential Purchase designation in Endocrinology Metabolic Disease Now fully revised and updated Williams Textbook of Endocrinology 15th Edition remains your go to reference for authoritative content on the full spectrum of adult and pediatric endocrine system disorders World renowned authors and editors expertly bridge the gap between basic science and clinical information keeping you up to date with recent advances in medications therapies clinical trials and more This essential reference is a must have resource for endocrinologists endocrine surgeons gynecologists internists pediatricians and other clinicians who

need current comprehensive coverage of this multifaceted field Presents current information in a highly illustrated user friendly format for quick reference Includes new chapters on Skeletal Regulation of Metabolism Digitized Approaches to Diabetes Therapeutics and MODY and Atypical Diabetes Reflects updated approaches to transgender medicine as well as new coverage of viral infections including COVID 19 Covers hot topics such as personalized medicine the latest methodologies and trends regarding cancer genomics precision oncology and cell biology and updates in key areas such as adrenal dysfunction and diabetes Provides state of the art coverage of diabetes metabolic syndrome metabolic bone disorders obesity and thyroid disease as well as pituitary gonadal and adrenal disorders and much more all designed to help you provide optimal care to every patient Features contributions from today s thought leaders in endocrinology Contains a selected reading list and highlighted key references nominated by the editors Research into Childhood-Onset Diabetes Andrea Scaramuzza, Carine de Beaufort, Ragnar Hanas, 2016-10-18 This book offers a detailed update on research into diabetes with onset in childhood or adolescence The coverage ranges from fundamental aspects such as study design and statistical analysis through to new ideas for research the latest research findings and implications of these findings for clinical care in childhood Examples of topics addressed in this context include the prevention of diabetic complications management of micro and macrovascular complications the roles of nutrition and exercise insulin therapy islet transplantation and risky behaviors in teenagers Attention is also focused on the latest technologies including the artificial pancreas and continuous glucose monitoring and associated regulatory aspects The reader will find novel insights into the ways in which research is impacting on clinical management as well as advice on methodology and stimulating ideas on future research areas The book is based on presentations delivered at the 15th ISPAD International Society for Pediatric and Adolescent Diabetes Research School for Physicians held in Milan It will be of interest for all diabetologists pediatricians dietitians biomedical engineers nurses and students The Underlying Mechanisms and Novel Approaches for Diabetes and its Related Complications Qi Pan, Lixin Guo, Chen Chen, Solomon Tesfaye, 2024-08-30 Diabetes mellitus is a chronic metabolic disease characterized by elevated blood glucose levels which severely affects the quality of life and survival of patients The latest data from the International Diabetes Federation showed that there are approximately 537 million diabetic patients globally in 2021 and it is expected to reach 783 million by 2045 The treatment regime of diabetic patients mainly refers to the management of blood glucose and its complications At present typical hypoglycemic medications include insulin and non insulin hypoglycemic drugs which can be subdivided into a glucosidase inhibitors metformin thiazolidinediones sulfonylureas glinides DPP 4i GLP 1RA and SGLT 2i Diabetic patients with poor islet function usually require insulin replacement but the majority of patients fail to meet ideal therapeutic targets with multiple hypoglycemic drug combinations including insulin Furthermore poor glycemic control is closely associated with an increased risk of diabetes complications consequently an increased rate of mortality More recently promising new drugs and therapies emerged which have many other important

benefits beyond glucose lowering In addition some traditional Chinese medicine have gradually revealed their therapeutic advantages All of them provide more choices for the treatment of diabetes patients **Endocrinology: Adult and** Pediatric E-Book J. Larry Jameson, Leslie J. De Groot, 2015-02-25 Considered the definitive source in its field for over 35 years Endocrinology Adult and Pediatric has been thoroughly updated to reflect today s recent advances in adult and pediatric endocrinology Unique perspectives from a team of trusted world renowned experts ensure this medical reference book remains the most highly regarded text in the field Make the best clinical decisions with an enhanced emphasis on evidence based practice and expert opinions on treatment strategies Zero in on the most relevant and useful references with the aid of a more focused concise bibliography Locate information guickly while still getting the complete coverage you expect Now in full color with special design treatment for at a glance pediatric content helping to distinguish the pediatric content Expanded coverage for key topics such as pediatric endocrinology and obesity mechanisms and treatment in addition to today s hot topics in endocrinology including endocrine disruptors bariatric surgery androgen deficiency genetic causes of obesity endocrine rhythms and the use of tyrosine kinase inhibitors in thyroid cancer New content addressing the latest advances in testosterone and estrogen replacement as well as the new causes of calcium and phosphate disorders new molecular causes of endocrine cancers new genetic causes of reproductive disorders and more Updated clinical guidelines for diabetes lipid disorders obesity management osteoporosis and more as well as essential treatment updates for the medical management of acromegaly Cushing's Disease hypercalcemia and diabetes mellitus New Key Points provide snapshots of what to expect in each chapter or serve as a refresher of what you just read Expert Consult eBook version included with purchase This enhanced eBook experience allows you to search all of the text figures references and videos from the book on a variety of devices Glucokinase and Glycemic Diseases Franz M. Matschinsky, Mark A. Magnuson, 2004 The glucose phosphorylating enzyme glucokinase also known as hexokinase IV plays a preeminent role as a glucose sensor and regulatory metabolic enzyme The implications of this central position of glucokinase for the understanding and treatment of diabetes mellitus are far reaching This book addresses the molecular and human genetic as well as the physiological chemical and pharmacological aspects of the role glucokinase plays in glucose homeostasis and diseases of glucose metabolism and how the enzyme serves as the drug target for the treatment of diabetes mellitus. The collection of assays by a team of internationally renowned authors includes basic science chapters dealing with the enzymology structure and the kinetics of hexokinases essays describing the biochemical genetics of glucokinase linked hypo and hyperglycemia syndromes and papers that discuss the diverse roles of the enzyme in a variety of tissues Finally the book contains the latest information on the recently discovered class of weight drugs that activate glucokinase i e glucokinase activators or GKAs This volume is of great interest to basic and clinical diabetes researchers geneticists biochemists physiologists and pharmacologists

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