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Neurometabolic Hereditary Diseases of Adults Alessandro P. Burlina,2018-06-04 This practical book describes only neurometabolic hereditary diseases which have a specific treatment and encourages the general neurologist to think of the most common neurometabolic hereditary diseases which he might have seen and never considered in the differential diagnosis Information regarding how to deal with diseases with special therapy is provided i e enzymatic replacement therapy in Fabry disease and Pompe disease as is information on diseases which are not easily recognized i e Niemann Pick disease type C and diseases with clinical features mimicking other common neurodegenerative diseases i e Wilson s disease Neurometabolic Hereditary Diseases is written with a clinical focus for adult neurologists working in general hospitals

Manual of Cytogenetics in Reproductive Biology Pankaj Talwar,2014-02-28 Cytogenetics is the study of the structure and function of the cell particularly chromosomes Manual of Cytogenetics in Reproductive Biology examines the diagnostic role of cytogenetics in improving the outcome of assisted reproductive technologies ART Divided into six sections the book begins with the basics of genetics followed by investigative cytogenetics applied cytogenetics recent advances preimplantation and prenatal cytogenetics This comprehensive guide includes nearly 200 clinical images diagrams and tables and is an invaluable reference for practising specialists in genetics infertility and obstetrics and gynaecology Key points Examines diagnostic role of cytogenetics in improving outcome of ART Six sections each providing in depth coverage of different aspects of cytogenetics Includes nearly 200 clinical images diagrams and tables Invaluable for specialists in genetics infertility and OBGY

Genomic management of animal genetic diversity J.K. Oldenbroek,2023-08-07 Recently developed genomic tools like SNP genotyping and whole genome sequencing and their analysis offer great opportunities for the conservation and utilisation of animal genetic diversity both among and within breeds These genomic tools can be used to detect potentially valuable rare alleles and haplotypes They are important parts of the genetic diversity we need to conserve now for possible utilisation in the future This book describes the use of genomic technology to define breeds to measure diversity and to assess important features in the history of breeds affecting the present genetic diversity The management of genetic diversity with genomic tools is outlined both in vivo small populations

of rare breeds or large populations with small effective population sizes and in vitro genebanks Special attention is given to the genomic management of populations of animals with high incidences of genetic defects This book is intended for MSc and PhD students scientists working with small populations in animal breeding and in conservation programmes for rare breeds

Color Atlas of Genetics Eberhard Passarge, 2017-12-13 Ever since the International Human Genome Project achieved its extraordinary goal of sequencing and mapping the entire human genome represented by approximately 3 billion base pairs with its far reaching implications for understanding the causes of human genetic disorders and their diagnosis progress in the field has not slowed down In the fifth edition of the bestselling Color Atlas of Genetics readers will be rewarded with a complete and current overview of the field with an emphasis on the interface between fundamental principles and practical applications in medicine and the role of signaling pathways in causing diseases Using the acclaimed Flexibook format designed for easy visual learning and retention the atlas is invaluable for students clinicians and scientists interested in staying up to date in this fast evolving area New fully illustrated topics in the revised fifth edition of the atlas include An overview of disorders resulting from structural changes of the genome genomic disorders Abnormal imprinting patterns Examples of impaired signal pathways laminopathies fibrillinopathies cohesinopathies and others The CRISPR Cas system Genetic features of the aging processes Disorders due to rearrangements of chromatin in the cell nucleus and others With almost 200 stunning color plates explained by concise texts on the opposite pages including tables presenting useful data a glossary of terms key references and online resources the atlas presents clear and accessible concepts It is an excellent refresher for investigators in any field of medicine or biology

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