Sexual Development
Genetics, Molecular Biology, Evolution, Endocrinology, Embryology and Pathology of Sex Determination and Differentiation
With the advent of affordable high-throughput genetic sequence, the human genetics community is returning to a focus on family data. Sequencing family members can facilitate data cleaning by identifying non-Mendelizing putative variants. It can provide a powerful mechanism for discerning the relevant sequence variants, by focusing on those segregating with the phenotype of interest. By the same token, sequencing holds out the possibility of capitalizing on earlier gene localization findings based on analyses of family data – findings that have provided numerous tantalizing loci for a wide range of complex phenotypes, but to date have resulted in relatively little definitive identifications of specific causal genes. This special issue of ‘Human Heredity’ is intended to showcase some of the advances in the statistical and computational technologies now available for linkage analysis. As the field returns to an appreciation of the power of linkage analysis, the collection of papers will convey the flavor of what is now possible. Human geneticists and scientists involved in gene mapping studies will gather information on new methods and computer programs available for linkage analysis to parallel sequencing experiments.

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Following technical and methodological improvements of the last decade, the underlying genes of a number of syndromes involving severe intellectual disability resembling Angelman and Rett syndromes have been identified. In order to keep track with these new entities, this special issue of Molecular Syndromology provides comprehensive reviews of the state of clinical and genetic knowledge about the old entities of Angelman and Rett syndromes, as well as the newer syndromes related to MECP2 duplication or defects in the CDKL5, FOXG1, MEF2C, TCF4, NRXN1, CNTNAP2, SHANK3, EHMT1 and FOXP1 genes. Furthermore the special challenge that presents itself when seeking to establish a diagnosis in adult patients is discussed. All articles are authored by experts specializing in these particular syndromes. This publication should therefore provide a unique source of knowledge about these relatively common syndromes and should be an asset to all clinical geneticists, neuropediatricians, and researchers in the field of neurodevelopmental disorders. Since this collection of articles is presented within a single issue, it facilitates comparison between the different syndromes and seems to be destined to become a desk book that anyone involved in the field of medical genetics dare not overlook.

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Monogenic Hyperinsulinemic Hypoglycemia Disorders

Charles A. Stanley
Diva D. De León

In this volume of Frontiers in Diabetes dealing with the molecular basis of monogenic disease of beta-cell insulin regulation, world-renowned experts provide in-depth descriptions of the many recent advances in genetic defects that cause hyperinsulinemic hypoglycemia. These disorders comprise the most important form of hypoglycemia in infants and children and are associated with a high risk of morbidity, including seizures and severe brain injury. The discovery of eight different genetic loci involved in congenital hyperinsulinism has led to greatly improved methods of diagnosis and treatment. New approaches to diagnosis are highlighted, such as 18F-DOPA PET scans for preoperative localization of focal hyperinsulinism, as well as potential new treatments, such as green tea polyphenols for GDH-HI and GLP-receptor antagonists for SUR1 and Kir6.2 hyperinsulinism. Practitioners, including pediatricians and specialists in endocrinology, surgery, genetics, pathology, and radiology, will find important up-to-date information for clinical diagnosis, management, and new treatments for infants and children with congenital hyperinsulinism. Researchers will discover how genetic hyperinsulinism disorders provide novel insights into the basic mechanisms regulating insulin secretion not only in diabetics, but also in healthy humans.

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The etiologies and pathologies of disorders of sex development of humans and animals explored by integrative approaches at the interface of several disciplines are relevant for fundamental biology as well as human and veterinary medicine. This special issue of Sexual Development presents expert reviews and previously unpublished research that detail the state-of-the-art understanding of the occurrence of abnormal genetic, gonadal and phenotypic sex of livestock and wild and captive-bred exotic animals. Providing insights into the fascinating and medically relevant causes and effects of disorders of sex development this issue is a valuable resource for researchers, clinicians as well as students of biology, animal production, agricultural sciences, human and veterinary medicine.

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