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Collana	JIMD Reports, , 2192-8304 ; ; 19
Disciplina	616.39042
Soggetti	Human genetics Metabolic diseases Pediatrics Human physiology Human Genetics Metabolic Diseases Human Physiology Genètica humana Trastorns del metabolisme Pediatria Fisiologia humana Llibres electrònics
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references at the end of each chapters.
Nota di contenuto	Contents; A De Novo Variant in Galactose-1-P Uridylyltransferase (GALT) Leading to Classic Galactosemia; Abstract; Introduction; Methods; Study Volunteers; GALT Genotyping of DNA from the Child; GALT Genotyping of DNA from Both Parents; Affymetrix 6.0 Microarray Analysis of Genomic Markers in the Trio; Results; Case Report; Dideoxy Sequencing Reveals a Novel GALT Variant in a Child with Classic Galactosemia; Dideoxy Sequencing of Parental GALT Loci Coupled with Comprehensive Genomic SNP Analysis of the Trio Reveals that c. 563A>C (p....; Discussion; 1-Sentence Synopsis

Compliance with Ethical GuidelinesConflict of Interest; Informed Consent; Animal Rights; Contributions of Each Author; References; Refsum Disease Presenting with a Late-Onset Leukodystrophy; Abstract; Discussion; One Sentence Take-Home Message; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Details of the Contributions of Individual Authors; References; Making the White Matter Matters: Progress in Understanding Canavans Disease and Therapeutic Interventions Through Eight Decad...; Abstract; Clinical Description
Aspartoacylase: Biochemistry and GeneticsCharacterization of the Substrate; Theories Behind the Molecular Etiology of CD; Molecular Water Pump (MWP) and Osmolyte Imbalance Theory; Dysmyelination Theory; Deficiency of AspA-Derived Acetate Compromises Oligodendrocyte Differentiation; Protein Folding and Stabilization Theory; Oxidative Stress Theory; Treatment Strategies; Palliative Measures; Symptomatic Treatment of Disease; Addressing Elevated Substrate in the Context of Deficient Aspartoacylase; Addressing the Deficiency of the Enzyme Aspartoacylase
Gene Therapy Using Gene Replacement StrategyPerspectives and Future Directions; One Sentence Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Details of the Contributions of Individual Authors; References; Disordered Eating and Body Esteem Among Individuals with Glycogen Storage Disease; Abstract; Introduction; Methods; Participants; Procedure; Measures; Eating Disorders Inventory-3 (EDI-3); Eating Disorders Inventory-Child (EDI-C); Eating Attitudes Test (EAT); Childrens Eating Attitude Test (ChEAT); Body Esteem Scale (BES)
Body Esteem Scale for Children (BES-C)Open-Ended Interview; Data Analysis; Results; Bullying/Teasing; Weight; Height; Negative Body Image; Positive Body Image; Age-Related Acceptance; Discussion; Conclusions; Compliance with Ethics Guidelines; Synopsis; Conflict of Interest; Informed Consent; Author Contributions; References; One Year Experience of Pheburane (Sodium Phenylbutyrate) Treatment in a Patient with Argininosuccinate Lyase Deficiency; Abstract; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Authors Contributions; References
Growth Hormone Deficiency and Lysinuric Protein Intolerance: Case Report and Review of the Literature

Sommario/riassunto

JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder, or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.