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Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	Contents; Growth Charts for Individuals with Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome); Abstract; Introduction; Methods; Data Source for Integrated Height Analysis; Definitions of Slowly and Rapidly Progressive Disease; Laboratory Assessments; Statistical Methods; Results; Demographics; MPS VI Growth Curves; Growth of Rapidly and Slowly Progressing MPS VI Patients; MPS VI Growth Charts and Z-Scores for Rapidly and Slowly Progressing MPS VI Patients; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Patient Consent Statement

Contributions of Individual AuthorsReferences; Treatment Adherence in Type 1 Hereditary Tyrosinaemia (HT1): A Mixed-Method Investigation into the Beliefs, Attitudes and Beha...; Abstract; Introduction; Methods; Sample; HCPs; Patients; Procedure; Qualitative Research; Quantitative Research; Results; Sample Characteristics; Psychological Well-Being and Social Support; Relationship with HCPs; Perceptions of Illness and Treatment; Adherence; Factors Associated with Adherence to Diet and Medication; Illness Experience and Perceptions; Treatment Perceptions and Experiences

Patient and Family FactorsDiscussion; Recommendations and Conclusions; Disclaimer; Source of Support; Take-Home Message; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; References; Regression of Hepatocellular Adenomas with Strict Dietary Therapy in Patients with Glycogen Storage Disease Type I; Abstract; Introduction; Methods; Results; Discussion; Take Home Message; Compliance with Ethics Guidelines; Conflict of Interest; Financial Disclosure; Other Relationships; Ethics Approval; Informed Consent; Animal Rights; Details of the Contributions of Individual Authors

ReferencesProteasome Inhibitor Bortezomib Enhances the Activity of Multiple Mutant Forms of Lysosomal -Glucosidase in Pompe Disease; Abstract; Introduction; Materials and Methods; Chemicals and Antibodies; Cell Cultures; Western Blot Analysis; Immunocytochemistry; Site-Directed Mutagenesis and Expression of Mutated GAA in HEK293T Cells; Quantitative Analyses of Proteins and GAA Activity; Results; Bortezomib Improves the Activity of Several Mutant GAAs in Fibroblasts from Patients with Pompe Disease

Bortezomib Improves the Maturation and Intracellular Localization of Mutant GAAs in Patient Fibroblasts Carrying M519V and C64... Bortezomib Increases the Activity of Several Mutant GAAs in HEK293T Cells; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest Disclosures; Informed Consent; Animal Rights; Contribution; References; Cognitive Function in Adults Aging with Fabry Disease: A Case-Control Feasibility Study Using Telephone-Based Assessments; Abstract; Introduction; Methods; Participants and Procedures; Participants with Fabry Disease; Control Participants

Measures

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.