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Nota di contenuto	JIMD Reports - Case and Research Reports, 2012/5; Contents; The Acid Sphingomyelinase Sequence Variant p.A487V Is Not Associated With Decreased Levels of Enzymatic Activity; Abstract; Introduction; Materials and Methods; Ethics Statement; Subjects; Genotyping; Overexpression of ASM Variants; Western Blot Analysis; In Vitro Determination of ASM Activity; Web Resources; Results; High Frequency of ASM Sequence Variation c.1460C>T in the General Population; Carriers of c.1460C>T Do Not Display Decreased Levels of ASM Activity ASM Variant p.A487V is Catalytically Active Upon Transient OverexpressionDiscussion; Author Contributions; References; The Changing Face of Infantile Pompe Disease: A Report of Five Patients from the UAE; Abstract; Introduction; Methods; Case Reports; Discussion; Conclusion; References; A Pilot Study of the Effect of (E, E) -2, 4-Undecadienal on the Offensive Odour of Trimethylamine; Abstract; Introduction; Methods; Preparation of the Study Reagents; Statistical Analysis; Results; Discussion; References; ALG6-CDG in South Africa: Genotype-Phenotype Description of Five Novel Patients;

## Abstract

IntroductionPatients and Methods; Patients; Blood/Serum Transferrin IEF; ALG6 Mutation Analysis; Results; Clinical and Biochemical Description of Patients; Blood/Serum Transferrin IEF; ALG6 Mutational Analysis; Discussion; Competing Interests; References; Unexplained Hypoglycemia During Continuous Nocturnal Gastric Drip-Feeding in a Patient with Glycogen Storage Disease Type Ia: Is It a Dumping-Like Syndrome?; Abstract; Introduction; Case Report; Discussion; Acknowledgments; References

A Dysmorphometric Analysis to Investigate Facial Phenotypic Signatures as a Foundation for Non-invasive Monitoring of Lysosomal Storage DisordersAbstract; Background; Methods; Ethics Approvals; Participants; 3D Image Acquisition; Anthropometric Masks and Facial Mapping; Statistical Face-Space; Dysmorphometrics and Normal Equivalents; Scoring, Analysis and Visualisation of Facial Variants; Normative Population Reference Statistics; Results; Discussion; Conclusions; Authors Information; Synopsis; Authors Contributions; Guarantor; Competing Interests; References

Orthotopic Liver Transplantation in an Adult with Cholesterol Ester Storage DiseaseAbstract; Introduction; Case History; Discussion; Synopsis; Conflicts of Interest and Financial Disclosures; References; Inheritance of the m.3243A>G mutation; Abstract; Introduction; Methods; Results; Discussion; Conflicts of Interest; Key Sentence/Synopsis; References; Recommendations on Reintroduction of Agalsidase Beta for Patients with Fabry Disease in Europe, Following a Period of Shortage; Abstract; Background; Methods; Results and Discussion; ERT Current Situation; Basic Principles of Treatment Criteria for Prioritization

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## 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.

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