CentoMD®: Genetic variants-related biomarker knowledge database

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Clinical symptoms in lysosomal storage diseases (LSDs) are caused by the deficiency of specific enzymes function and resultant substrate accumulation in the lysosomes. Several biomarkers are already in use as indicators of the presence and monitoring of LSDs: lyso-Gb3 (Gb3) in Fabry disease (FD), lyso-Gb1 (Gb1) in Gaucher disease (GD) and NP509 in Niemann-Pick (NP) disease. CentoMD® is a browser-based tool that enables access to a high-quality repository of genetic, biochemical and human phenotype ontology (HPO)-based clinical information. All patients provided informed consent before inclusion in the DB. We measured lyso-Gb3, lyso-Gb1 and NP509 in the DBS samples obtained from 5,603 patients (57.6% females, 39.1% males, 3.3% unknown) undergoing biochemical and genetic testing for verification of FD (71.7%), GD (15.8%) or NP (12.5%). The pathological cut-off for biomarker measurements was set to 1.8 ng/ml for Gb3, to 4.8 ng/ml for Gb1; and to 0.9 ng/ml for NP509. Biomarker levels were correlated with clinical severity of the individual patients.