Abstracts
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Novel and Known Morbidities of Leukodystrophies Identified Using a Phenome-Wide Association Study

Objective To determine shared comorbidities and to identify underrecognized or unexpected morbidities in children with leukodystrophies using an unbiased phenome-wide association study (PheWAS) analysis of a nationwide pediatric clinical and financial database.

Methods Data were extracted from the Pediatric Health Information System database. Patients with leukodystrophy were identified with International Classification of Diseases, 10th revision, clinical modification, diagnostic codes for any of 4 specific leukodystrophies (X-linked adrenoleukodystrophy [E71.52x], Hurler disease [E76.01], Krabbe disease [E75.23], and metachromatic leukodystrophy [E75.25]) over a 3-year time period. Confirmed leukodystrophy cases (n = 553) were matched with 1,659 controls. A PheWAS analysis was performed on all available ICD diagnostic codes for cases and controls. Comparisons were performed for all 4 leukodystrophies as a group and individually.

Results We found 174 phecodes (grouped ICD codes) associated with leukodystrophies, including 28 codes with a rate difference (RD) >20%. Known comorbidities of leukodystrophies including developmental delay, epilepsy, and adrenal insufficiency were identified. Unexpected associations identified included hypertension (RD 30%, OR 25), hearing loss (RD 28%, OR 15), and cardiac dysrhythmias (RD 27%, OR 9). Hurler disease had a greater number of unique disease conditions.

Conclusions PheWAS analysis from a national database demonstrates shared and unique features of leukodystrophies. Developmental delay, cardiac dysrhythmias, fluid and electrolyte disturbances, and respiratory issues were common to all 4 leukodystrophy diseases. Use of a PheWAS in leukodystrophies and other pediatric neurologic diseases offers a method for targeting improved care for patients by identification of morbidities.

Cryptogenic Stroke: Contemporary Trends, Treatments, and Outcomes in the United States

Background Nationwide data on patients with cryptogenic stroke (CS) are lacking. We evaluated patient and hospital characteristics, in-hospital treatments, and discharge outcomes among patients with CS compared with other subtypes in the Get With The Guidelines (GWTG)-Stroke registry.

Methods We identified patients with ischemic stroke (IS) admitted to GWTG-Stroke participating hospitals between January 1, 2016, and September 30, 2017, with documented NIH Stroke Scale (NIHSS) scale and stroke etiology (cardioembolic [CE], large artery atherosclerosis [LAA], small vessel occlusion [SVO], other determined etiology [OTH], or CS). Using multivariable logistic regression, we compared hospital treatments and discharge outcomes by subtype, adjusted for patient and hospital characteristics.

Results Among 316,623 patients from 1,687 hospitals, there were 63,301 (20.0%) patients with CS. In multivariable analysis, patients with CS received IV thrombolysis more often than other subtypes and had lower mortality than CE, LAA, and OTH but higher mortality than SVO. They were more likely to be discharged home than all other subtypes and be independent at discharge than LAA, OTH, or SVO.

Conclusions In a large contemporary nationwide registry, CS accounted for 20% of ISs among patients with a documented stroke etiology. Patients with CS had a distinct profile of treatments and outcomes relative to other subtypes. Improved subtype documentation and further research into CS are warranted to improve care and outcomes for patients with stroke.

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