

electronic data warehouse (EDW) and then matched to records in CHND. With severity of HIE, gender, and confirmed seizures, each marker's association with LOS was calculated using multivariable Cox proportional hazards regression equations. These analyses were stratified by mortality. Candidate markers were vital signs, pulse oximetry, creatinine, acidosis (pH), international normalized ratio (INR), and supplemental oxygen (FiO₂). RESULTS/ANTICIPATED RESULTS: There were 66 eligible infants (38 males) and 1741 patient-days identified; Severe HIE (48%) and mortality (n = 21, 32%) were common. Overall, the median length of stay (mLOS) was 20.5 days (25th–75th centile: 10–31 days), although shorter for nonsurvivors [nonsurvivors mLOS = 8 days (5, 20); survivors mLOS = 24 days (14, 31), $p < 0.001$]. Median birthweight and gestational age were 3.3 kg and 39.4 weeks' gestation, respectively. In survivors (n = 45, 1290 days), regression analyses demonstrated that none of the selected parameters were associated with LOS. Among nonsurvivors (n = 21, 451 days), diastolic blood pressure changes [hazard ratio (HR) = 0.93, 95% confidence interval (CI) = 0.88, 0.97, $p = 0.04$] was related to longer time of survival; conversely, temperature (HR = 2.0, 95% CI = 1.24, 3.26, $p = 0.005$) was related to shorter survival. Creatinine, pH, INR, FiO₂, or other vital signs were unrelated to time-to-death in nonsurvivors. DISCUSSION/SIGNIFICANCE OF IMPACT: In a pilot study of neonatal HIE, changes in physiologic values were related to duration of survival in nonsurvivors, while neither physiologic nor laboratory values were related to survivors' mLOS. These results both exemplify novel uses for disease-specific, exposure-outcome relationships using EDWs and incorporates required functionalities of required software patches to extract, clean, and report from clinical information captured in electronic health records. We anticipate that text mining with techniques such as natural language processing will augment associations and/or predictions of short-term outcomes.

2240

High-throughput phenotyping and the increased risk of OSA in Rosacea patients

Peter Elkin, Sarah Mullin, Sanjay Sethi, Shyamashree Sinha and Animesh Sinha

University at Buffalo, State University of New York, Buffalo, NY, USA

OBJECTIVES/SPECIFIC AIMS: To create a new semantically correct high-throughput phenotyping (HTP) platform. To demonstrate the utility of the HTP platform for observational research and can allow clinical investigators to perform studies in 5 minutes. To demonstrate the improved accuracy of observational research using this platform when compared with traditional observational research methods. To demonstrate that patients who have Rosacea are at increased risk of having obstructive sleep apnea (OSA). METHODS/STUDY POPULATION: This population is a set of 212,343 patients in the outpatient setting cared for in the Buffalo area over a 6-year period. All records for these patients were included in the study. Structured data was imported into an OMOP (OHDSI) database and all of the notes and reports were parsed by our HTP system which produces SNOMED CT codes. Each code is designated as a positive, negative or uncertain assertion and compositional expressions are automatically generated. We store the codified data 750,000,000 codes in Berkeley DB, a NOSQL database, and we keep the compositional graphs in both Neo4j and in GraphDB (a triple store). Labs are coded in LOINC and drugs using RxNorm. We have developed a Web interface in .Net named BMI Search, which allows real-time query by subject matter experts. We analyzed the accuracy of structured Versus unstructured data by identifying NVAF cases with ICD9 codes and then looked for any additional cases based on the SNOMED CT encodings of the clinical record. This was validated by 2 clinical human review of a set of 300 randomly selected cases. Separately we ran a study to determine the relative risk of OSA with and without Rosacea using the data set described above. We compared the rates using a Pearson χ^2 test. RESULTS/ANTICIPATED RESULTS: We are able to parse 7,000,000 records in an hour and a half on 1 node with 4 CPUs. This yielded 750,000,000 SNOMED CT codes. The HTP data set yielded 1849 cases using ICD9 codes and another 873 using the HTP-NLU data, leading to a final data set of 2722 cases from our population of 212,343 patients. In total, 580 patients had Rosacea; 5443 patients had OSA without Rosacea and 51 patients had OSA with Rosacea. Patients with Rosacea had an 8.8% risk of OSA whereas patients without Rosacea only had a 2.6% risk of OSA. This was highly statistically significant with a $p < 0.0001$ (Pearson χ^2 test). The number needed to test was only 12. DISCUSSION/SIGNIFICANCE OF IMPACT: HTP can change how we do observational research and can lead to more accurate and more prolific investigation. This rapid turn around is part of what is necessary for both precision medicine and to create a learning health system. Patients with Rosacea are at increased risk of and should be screened for OSA.

2246

Characterization of resistant hypertension in a statewide electronic health record-based database (OneFlorida)

Caitrin W. McDonough, William R. Hogan, Betsy Shenkman and Rhonda M. Cooper-DeHoff

OBJECTIVES/SPECIFIC AIMS: Our objective is to create a Resistant Hypertension (RHTN) computable phenotype from electronic health record (EHR)-based data, and to determine the characteristics associated with RHTN within a large, diverse, EHR-based database. METHODS/STUDY POPULATION: The OneFlorida Clinical Research Consortium includes 10 unique health care systems providing care for approximately half of the state (48%, ~10 million). OneFlorida houses a Data Trust which contains longitudinal EHR data and claims data from these providers in a common format, the PCORnet common data model v3.0. For the current project, data from 5 health care systems were considered. All of the adult hypertension (HTN) patients with a HTN diagnosis from an outpatient encounter were extracted from the OneFlorida Data Trust. Additional data such as demographics, prescribing, and vitals information were also extracted. The RHTN computable phenotype was created by constructing a drug exposure variable that took into consideration the number of antihypertensive medications an individual was prescribed at any point in time over the course of the OneFlorida dataset. RHTN was defined as any blood pressure requiring four or more antihypertensive drugs, or uncontrolled blood pressure ($\geq 140/90$) on 3 antihypertensive drugs. RHTN cases had to meet the definition criteria twice during the data period, at least 30 days apart. All data extraction, computation phenotype coding, and statistical analyses were conducted using SQL or SAS. RESULTS/ANTICIPATED RESULTS: Our preliminary results show that there were n = 342,026 adults with a HTN diagnosis from an outpatient visit in the data set. After the RHTN computable phenotype was constructed, n = 11,670 RHTN cases were identified from the n = 130,901 HTN individuals with all of the required variables in the data set (8.9% RHTN prevalence). In all, 55% of RHTN cases were Black or African American, compared with the total HTN population (25% Black/African American). RHTN cases also had more prescriptions for loop diuretics, centrally acting agents, α -blockers, and vasodilators compared with the total HTN population. Not surprisingly, the RHTN cases had 26% of the antihypertensive prescriptions in the data set, and the RHTN cases had fewer blood pressure readings that were in control (only 49.4% of readings $< 140/90$). DISCUSSION/SIGNIFICANCE OF IMPACT: Overall, our preliminary data shows that it is possible to create the very complicated computable phenotype of RHTN within the OneFlorida Data Trust. We found that the RHTN prevalence in OneFlorida is 8.9% which is consistent with previous studies from NHANES. Although promising, these results require further validation of the computable phenotype and replication in other similar data sets in order to ascertain their true meaning. Once validated, the experience gained from this computable phenotype can be applied to many other phenotypes.

2278

Identifying causative mutations in Treacher Collins syndrome using iobio

Alistair N. Ward, Matt Velinder, Chase Miller, Tony Di Sera, Yi Qiao, Dave Viskochil and Gabor Marth

The University of Utah School of Medicine, Salt Lake City, UT, USA

OBJECTIVES/SPECIFIC AIMS: The objective of the study was 2-fold; to identify potentially deleterious alleles in a child with Treacher Collins syndrome, and; to demonstrate the value of the iobio analysis platform for intuitively and rapidly analyzing genomic data. METHODS/STUDY POPULATION: We used the iobio suite of web-based applications to analyze quality metrics for the sequencing data and called variants for the proband and his parents. We then visually interrogated variants in genes potentially associated with the syndrome in real-time, using the intuitive gene.iobio application. We sought high impact variants that demonstrated a predicted impact on the protein function, and were simultaneously at low allele frequency in the general human population. Variants were also compared against the ClinVar database of known mutations to identify variants that have already been associated with this, or related syndromes in the literature or clinical studies. Finally, the gene.iobio tool allows users to interrogate the primary sequencing data to ensure that no variants had been missed by the primary variant calling pipeline. This analysis pipeline was performed using intuitive web-based apps in real time, and consequently represents a system that is available to users that traditionally are excluded from these analyses. RESULTS/ANTICIPATED RESULTS: The iobio suite was used to rapidly assess data quality and interrogate genetic variants for a child with