Panel 2: Discovery Science and the Electronic Health Record

National Human Genome Research Institute

National Institutes of Health

U.S. Department of Health and Human Services

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National Institutes of Health

National Human Genome Research Institute

Genomics and Health IT Systems

April 27, 2011

"Electronic" Phenotyping

Library of Phenotype Algorithms

1 Cardiac Conduction

2 Cataract

3 Dementia

4 Diabetic Retinopathy

5 HDL

6 Height

7 Hypothyroidism (Presumptive Hashitomo's thyroiditis)

8 Lipids

9 PAD

10 Phenome-Wide Association Studies (PheWAS)

11 Red Blood Cell Indices

12 Type II Diabetes

13 White Blood Cell Indices

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https://www.mc.vanderbilt.edu/victr/dcc/projects/acc/index.php/Main_Page

"Where Electronic Medical Records and Genomics Meet"

REPORT

ELECTRONIC MEDICAL RECORDS

The investigators show that even though the EMRs were of different types and did not all use NLP… they were able to obtain robust positive and negative values for identifying patients with these diseases with sufficient accuracy for use in GWAS. They conclude that widespread adoption of EMRs will provide real-world clinical data that will be valuable for GWAS and other types of genetic research.

Clinical ... for re-search. ... ptured through ... gative predictive values for use in genome-wide association studies (GWAS). Using data from five different sets of EMRs, we have identified five disease phenotypes with positive predictive values of 73 to 98% and negative predictive values of 98 to 100%. Most EMRs captured key information (diagnoses, medications, laboratory tests) used to define phenotypes in a structured format. We identified natural language processing as an important tool to improve case identification rates. Efforts and incentives to increase the implementation of interoperable EMRs will markedly improve the availability of clinical data for genomics research.

21 Research Domains

Alcohol, Tobacco	Ocular
Anthropometrics	Oral Health
Cancer	Physical Activity/Fitness
Cardiovascular	Psychiatric
Demographics	Psychosocial
Diabetes	Reproductive Health
Environ. Exposures	Respiratory
Gastrointestinal	Skin, Bone, Muscle
ID /Immunity	Social Enviroments
Neurology	Speech/Hearing
Nutrition	

https://www.phenx.org/