

## 2<sup>nd</sup> International Conference on **Predictive, Preventive and Personalized Medicine & Molecular Diagnostics**

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## Pharmacogenomics at Boston Children's Hospital

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The overall goal of clinical and research pharmacogenomics (PGx) at Boston Children's Hospital (BCH) is to ultimately L improve medication safety through precision genetic medicine. The Clinical Pharmacogenomics Service (CPS) was created to facilitate the incorporation of pharmacogenomic (PGx) information into the medication management cycle. The service provides consultations for the clinicians at BCH who request assistance with the interpretation and application of relevant PGx data for patient care. Complete with an Oversight Committee composed of clinical experts who ensure the safe and rational movement of drug/gene pairs and return of results to the electronic medical record, the CPS is also responsible for the creation, maintenance and monitoring of related decision support rules in the EMR. This has highlighted potential problem areas in the clinical application of PGx, such as overcalling secondary to the inability to resolve diplotypes, and the ability to convey uncertainty in a meaningful way to patients when dealing with difficult to interpret regions. Careful metrics are maintained, such as clinical decision support statistics, outcomes, and adverse drug reaction (ADR) avoidance. Early statistics related to cost-savings from ADR avoidance have been generated. In the coming months, the CPS aims to evolve into a billable service with clinic hours. Research in pharmacogenomics at BCH is also thriving. Educational sessions have been developed in response to needs identified. BCH is also retrospectively studying the relationship between genotype and reported ADRs. One exciting project is enrolling patients from participating clinics (epilepsy, inflammatory bowel disease, renal transplant) and genotyping them on a broad PGx platform, returning results to providers and a selected subset of results to the EMR. This project also assesses the patient's response to medication and outcome in the context of their genotype and phenotype. Finally, groundbreaking research out of two Harvard laboratories has created a novel all-optical electrophysiology platform to rapidly screen drugs for functional effects in human neurons and cardiomyocytes. BCH considers clinical and research pharmacogenomics essential to offering world-class patient care using state-of-the-art drug/gene knowledge.

## Biography

Catherine Brownstein is a geneticist and toxicologist with eleven years experience in human genetics and three years in applying patient-reported outcomes to the study of disease. She has been served as a Project Manager of the Research Sequencing Program at Boston Children's Hospital since 2011. Her research career includes training in Genetics, Medical Genetics, Epidemiology, and Environmental Health. She has also completed my MPH at the Yale School of Epidemiology and Public Health and worked as a toxicologist at the Massachusetts Department of Public Health. Before coming to BCH and HMS, she spent four years creating online patient communities for individuals with chronic or terminal diseases.

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