

DRAFT MEETING SUMMARY
Unifying the Evaluation and Implementation of Genomic Medicine
August 18, 2016, Bethesda, MD

On August 18th, 2016, IGNITE convened a meeting entitled “Unifying the Evaluation and Implementation of Genomic Medicine” to address issues related to insurance coverage and access to genomic medicine. The meeting had the following objectives:

- To begin to build a process for communication among patients, providers, insurers, and researchers for a team-oriented approach to evaluating and implementing genomic medicine
- To understand what evidence is needed and how it should be disseminated for all
- To identify protocols that will help to provide evidence needed to make the application of genomic medicine sustainable

The meeting, including its objectives and agenda, were developed during several months of planning via weekly conference calls by a committee headed by Toni Pollin, Ph.D., University of Maryland IGNITE PI and then Steering Committee Chair and consisting of investigators and NHGRI program staff from IGNITE and other NHGRI genomic medicine programs (CSER, NSIGHT, eMERGE, and UDN) as well as a payer representative (Deborah Smith, MD, MPH, Director Blue Cross/ Blue Shield Federal Employee Program), two health economists [Daniel Mullins, PhD (University of Maryland School of Pharmacy) and Kathryn Phillips, PhD (University of California San Francisco)].

There were 66 in-person attendees and 36 remote attendees (via WebEx). Attendees/speakers included members and program staff, of IGNITE and other NHGRI genomic medicine networks and genetics organization, two patient advocates, in vitro diagnostics company representatives, NSGC President Jehannine Austin, PhD, geneticists, genetic counselors, health care providers, and six payer organization consultants:

- Suzanne Belinson, PhD (BlueCross BlueShield [BCBS])
- Joshua Plavin, MD (BCBS Vermont)
- Cheryl Reid, MD (Aetna)
- Kenneth Schaecher, MD (SelectHealth)
- Heather Shappell, MS, CGC (Beacon Laboratory Benefit Solutions)
- Deborah Smith, MD, MPH (BCBS Federal Employee Program)

The meeting opened with introductions by the NHGRI Director Eric Green and planning committee chair and co-chair Toni Pollin, PhD, and Daniel Mullins, PhD, a keynote speech by John Brumsted, MD, Chief Executive Officer of the University of Vermont (UVM) Medical Center and The UVM Health Network, and Joshua Plavin, MD, Senior Medical Director, BCBS Vermont.

Meeting activities focused on payer-moderated discussions around three briefly presented case studies of examples of genomic implementation involving genetic testing of increasing complexity and varying stage of implementation, presented from both the medical/scientific and consumer perspectives:

- Targeted genotyping (CYP2C19 variants in patients undergoing cardiac catheterization)

As of 5/1/17

- Targeted sequencing panel-based genetic testing (Diagnosing highly penetrant forms of diabetes)
- Genome-wide methods (arrays, exome, genome: Implementing exome sequencing for developmental delay and Vermont statewide genomic sequencing plan)

There was also a working lunch whereby 10-12 people convened at one of six roundtables to address targeted questions around the following topics:

- Building a coalition
- Evidence needed
- Designing research and clinical protocols to provide evidence
- Disseminating the evidence

The meeting was a forum for engaging the various stakeholders in delineating the challenges faced in implementing advances in genomic medicine into a complex health care system.

Challenges/goals highlighted include:

- The lack of coverage models for human and bioinformatics resources around communication and explanation of results in addition to cost of testing
- The lack of coverage models for re-analysis of results from genomic tests (including not just previously identified variants and genes but additional genes as in the case of whole genome sequencing) and transporting those results when patients move into a different healthcare system, particularly as universal genome sequencing becomes closer to reality
- Challenges in developing economic models that account for benefits and outcomes of genomic testing outside the purview of the entities providing care to the patient, including testing of family members, family planning, and getting a diagnosis to inform educational plans for children with special needs
- The need for models for providing updated information on genomic service utility to payers
- The need for specificity of CPT codes so it is clear which test is being performed
- Role and value of whole genome sequence as an ongoing resource to inform diagnosis and treatment
- Defining clinical utility beyond effects on medical management
- Genomic panel standardization

There was agreement that this meeting is to be the beginning of an ongoing strategic process for engaging diverse stakeholders in clarifying how to implement and sustain genomic medicine in the context of ongoing changes in testing methods and a complex healthcare system.

Expected deliverables in the next six months include:

- Meeting proceedings
- Manuscript based on meeting discussions
- A strategic plan for ongoing efforts to engage stakeholders and address challenges and obtaining resources to support it