"Peer-to-Peer": A program to Connect a Clinical Genomicist and the Treating Neurologist to Correlate Genotype with Phenotype
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Objective: To introduce and assess a model that utilizes telemedicine consultations between a clinical genomicist and the treating neurologist to understand and apply genetic information to alter and improve treatment.

Background:
The recent explosion in genetics has left clinicians unaware or confused in regards to how to interpret genetic testing. As testing finds increasing clinical utility among typical cases, the common approach of referral to research institutions faces increasing obstacles. For neurologists practicing outside these institutions, such referral burdens families, who may not return to the practice. The common result is inadequate quality and quantity of testing.

Currently, a main obstacle to routine genetic testing is the correlation of the thousands of genomics variants identified with the disease process in each unique patient. This requires an in-depth knowledge regarding genomics, which few clinicians possess, and of the individual patient, which testing laboratories cannot possess.

Design/Methods: The “Peer-to-Peer” Service connects a clinical genomicist with the treating neurologist. Through a telemedicine encounter, the patient and DNA sequences are presented, followed by a discussion regarding the meaning and utility of variants thought to be disease associated. The emphasis is on “actionable” findings that alter management. The genomicist is also available pre-testing to answer questions regarding whom should be tested, and by which tests.

Results: To date, 8 treating physicians in 7 states are enrolled. Seventeen patients have been evaluated, with tests assayed in 6 different genetics laboratories. Variants of clinical interest that altered the physicians’ management were identified in 16/17 cases, and a diagnosis was provided in 9/17. Furthermore, changes were recommended in therapy (15/17), additional testing (7/17), and outside referral (4/17).

Conclusions: Our early experiences have supported the viability of this model, and physician and family satisfaction have been excellent. We are collecting additional data, including outcomes of suggested testing and therapies.