A Genetic-Counseling-First, Telehealth Service Delivery Model for Pediatric Patients

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INTRO

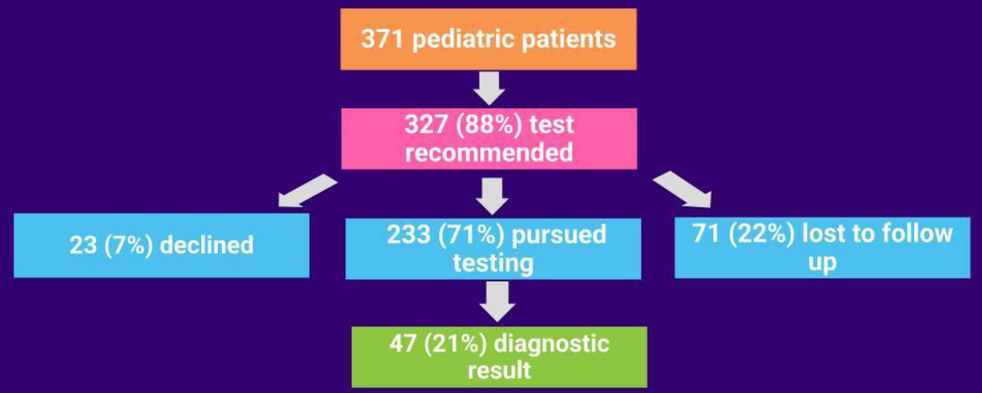
- Wait times for access to pediatric geneticists have increased, as the number of geneticists has not kept pace with increased demand for access
- Wait times for a pediatric geneticist are typically at least 3 months (Jenkins, *et al.*), though some clinics can reach up to over a year
- Medical genetics clinics are also mainly clustered in urban areas which create accessibility issues to those families in rural areas
- Because of these barriers to access we sought to determine if a genetic-counseling-first approach via our nationwide telehealth clinic would assist in either diagnosing or obtaining initial testing prior to a medical genetics referral for pediatric patients

METHODS

- We identified patients under the age of 18 seen by our genetic counseling practice over the last two years and queried:
  - Referral source
  - Indication
  - Testing recommendations
  - Testing outcomes
- Based on the above information, we sought to determine if our approach helped in:
  - Diagnosis of the patient with positive genetic testing
  - Referral back to genetics for additional testing and in person physical exam
  - Release from further genetic follow up pending any new concerns based on:
    - Negative testing and/or non concerning personal/family history
    - Diagnosis made not needing additional genetic evaluation (e.g. managed by neurology)

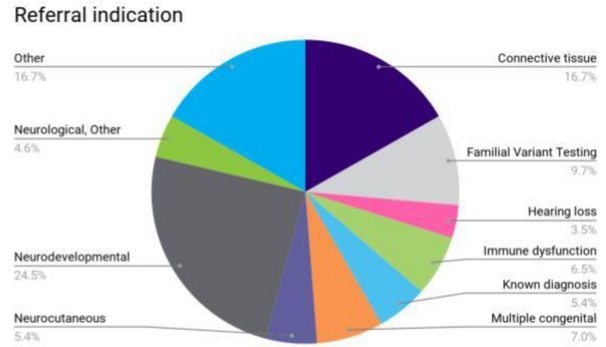
53% of pediatric patients seen through telehealth, GC first approach were able to be resolved without recommendations for additional in person genetics evaluation. Thus, this model could help reduce the burden for medical genetics departments.

Test Result	Referred back to genetics for additional evaluation	Resolved by genetic counselor
Positive (n = 47)	12 (25.5%)	18 (38.2%)
Negative (n = 179)	40 (15%)	106 (59.2%)
Not Recommended (n = 44)	5 (11.3%)	18 (40.9%)
Total (n = 270)	**57 (21%)**	**142 (52.6%)**



RESULTS:

- A total of 371 patients were identified with an average age of 7.9 months (range 1 mo - 18 years)
- 48% of patients were self referred and 52% were referred by a physician
- A total of 142 were able to be resolved by genetic counselor with no additional in person genetics evaluation recommended



DISCUSSION

- This model highlights the utility of a genetic-counseling-first approach as a triage and initial assessment step
- The early application of genetic testing for those patients unable to quickly be evaluated by a medical geneticist has a demonstrable yield, both in achieving diagnoses and in resolving pending cases to be seen by the geneticist
- This model could be utilized to help reduce waitlist times for medical genetics

genome MEDICAL