Improvement of Developmental Delay, Autism, and Seizure Diagnosis Before and After Exome Screening

ECU HEALTH | ECU BRODY SCHOOL OF MEDICINE

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BACKGROUND

- Developmental delays, autism spectrum disorders, and generalized seizures are common pediatric conditions that often have underlying genetic causes. Genetic disorders affect approximately 1 in 20 individuals globally, with many conditions manifesting in childhood.
- Despite their prevalence, these conditions frequently lead to diagnostic odysseys that delay critical interventions. Traditional diagnostic methods may fail to identify the genetic underpinnings, leaving patients and families without clear answers or targeted treatment options.

PROJECT AIM

At ECU Health, the growing pediatric population with complex presentations underscores the urgent need for precise diagnostics. By integrating exome sequencing with medical genetics consultations, we aim to increase diagnostic accuracy by 50% within 12 months for patients presenting with developmental delays, moderate to severe autism, or generalized seizures. This initiative not only addresses a critical gap in care but also advances precision medicine, providing children with the timely and targeted interventions they deserve.

PROJECT DESIGN/STRATEGY

Study Design

•**Baseline Data Collection (Pre-Intervention)**:
- Review patient records prior to November 2023 to assess diagnostic rates and timeframes without genetic consultations or exome testing.

•**Post-Intervention Monitoring**:
- Track all patients receiving genetic consultations and exome testing.
- Record diagnostic outcomes and timeframes for comparison with baseline data.

•**Outcome Measures:**
- Review patient records prior to November 2023 to assess diagnostic rates and timeframes without genetic consultations or exome testing.

•**Post-Intervention Monitoring**:
- Track all patients receiving genetic consultations and exome testing.
- Record diagnostic outcomes and timeframes for comparison with baseline data.

CHANGES MADE (PDSA CYCLES)

1
- Establish genetics referral and testing workflow
- Train staff on new protocols for identifying and referring eligible patients
- Monitor adherence to referral criteria and process efficiency

2
- Optimize Testing and Follow-up
- Evaluate time from patient presentation to test results and confirmed diagnosis
- Identify and address delays or barriers in the process

3
- Use secured electronic health records to track patient data
- Categorize patients based on: immediate, delayed, or absent genetic consultations/testing and diagnostic outcomes (confirmed genetic conditions
- Analyze trends in diagnostic accuracy and timeliness
- Compare post-intervention outcomes to baseline data; prepare final report

Target Population

•Pediatric patients at ECU Health presenting with:
- Persistent or global developmental delays
- Moderate to severe autism spectrum disorder
- Generalized seizures

•Focus on patients with unexplained symptoms that meet criteria for genetic testing.

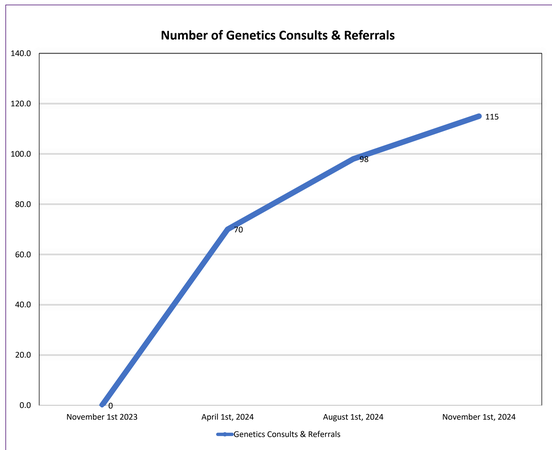
Intervention

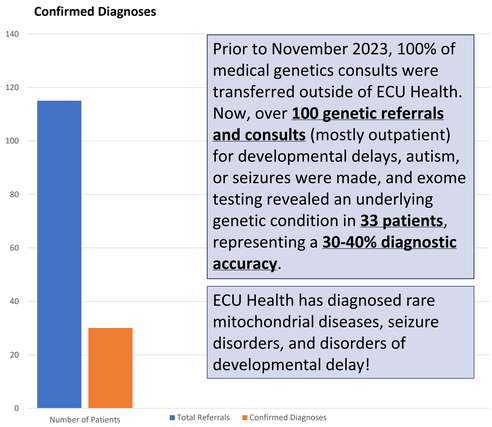
•Implement exome and genome sequencing as a routine diagnostic tool for eligible patients.

•Provide immediate genetic consultations upon patient presentation with qualifying symptoms.

•Develop protocols for timely referrals and testing:
- Establish criteria for triggering genetic referrals.
- Ensure a streamlined process from presentation to consultation and testing.

RESULTS/OUTCOMES





Prior to November 2023, 100% of medical genetics consults were transferred outside of ECU Health. Now, over **100 genetic referrals and consults** (mostly outpatient) for developmental delays, autism, or seizures were made, and exome testing revealed an underlying genetic condition in **33 patients**, representing a **30-40% diagnostic accuracy**.

ECU Health has diagnosed rare mitochondrial diseases, seizure disorders, and disorders of developmental delay!

LESSONS LEARNED

- Exome and Genome Sequencing can reduce diagnostic odysseys for complex pediatric cases via earlier identification of genetic conditions
- Earlier identification of genetic conditions
- Multidisciplinary collaboration between pediatric teams, medical geneticists, and laboratory services to implement an effective workflow
- Standardized workflows reduce delays and ensure eligible patients are promptly tested
- Identifying bottlenecks (e.g. delays in referrals or testing) enables targeted interventions to address gaps

NEXT STEPS

Expand Genetic Testing Accessibility
- Broadening testing criteria
- Improving access to genetic testing to patients for rural or underserved Eastern NC

Develop Comprehensive Education Programs
- Provide ongoing training on the indications, benefits, and limitations of exome sequencing to ensure appropriate utilization

Enhance Workflow Efficiency
- Use automated EHR alerts to identify patients who meet genetic testing criteria

Improve Multidisciplinary Collaboration
- Enhance coordination between primary care providers, geneticists, and other specialists to ensure continuity of care

ACKNOWLEDGEMENTS

Thank you to all the ECU Health providers, medical, and support staff that have helped integrate this initiative. And most importantly, we would like to thank our patients and their families for whom we wish to continue to serve.

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