New Frontier: The First Year of an Adult Neurodevelopmental Disabilities Clinic

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Purpose

1. Characterize the patient population seen in the first year of a newly developed Adult Neurodevelopmental Disabilities (NDD) Clinic.
2. Demonstrate yield of genetic testing in adults with neurodevelopmental disabilities (NDD)

Background

More than 7 million individuals in the United States live with a neurodevelopmental disorder (NDD). Despite the increasing need for high-quality care for these individuals, few physicians specialize in caring for adults with NDDs, and care often lacks coordination among health care services. The lack of coordination and paucity of skilled services in all life stages can lead to significant health disparities. As patients with NDD transition from pediatric to adult healthcare systems, they often have difficulty finding providers who address their NDD-related needs, including revisiting the etiologic workup of their NDD. In response to this care, a new consultation service for high-quality care for these individuals, few physicians specialize in caring for adults with NDDs, and care often lacks coordination among health care services. The lack of coordination and paucity of skilled services in all life stages can lead to significant health disparities. As patients with NDD transition from pediatric to adult healthcare systems, they often have difficulty finding providers who address their NDD-related needs, including revisiting the etiologic workup of their NDD. In response to this care, a new consultation clinic was established in an adult neurology department to address neurodevelopmental concerns of these adult patients and provide etiologic workup, including genetic testing.

Methods

Data was obtained by a retrospective chart review of all 86 patients seen in the adult Neurodevelopmental Disabilities clinic from September 2020 through December 2021.

Results

Developmental diagnoses included but were not limited to:
- Autism Spectrum Disorder (47%)
- Intellectual Disability (63%), Down syndrome (15%)
- Cerebral Palsy (9%) and other genetic disorders (26%)

Comorbidities addressed included:
- Anxiety (29% of patients), behavioral concern (34%), seizures (22%), and depression (15%)

New genetic testing was completed in 16 patients.
- Overall yield: 11/16 (69%)
- Chromosomal Microarray (CMA) in 3/11 (27%)
- Autism Spectrum Disorder (ASD)/Intelligence Disability (ID) panel in 3/10 (30%)
- Whole Exome Sequencing (WES) in 1/2 (50%)

Conclusion

Our study characterized the diversity of developmental disabilities and comorbidities addressed in a new specialty Adult NDD clinic. This retrospective report of the first year of a new adult NDD clinic demonstrates both the need for and feasibility of, serving this population within an adult neurology setting.

Reference


Clinical Relevance

So far, 11 adults received genetic explanations of their neurodevelopmental disorders. All 11 of the patients’ genetic testing influenced plan of care and/or further surveillance.

Discussion

The study describes a clinic that specializes in the complex neurodevelopmental care of patients with NDD. A variety of patients with different developmental diagnoses, genetic diagnoses, and demographics were seen in the first year of the adult NDD clinic. The clinic’s diversity and long wait list highlights the growing need for specialized adult neurodevelopmental clinics. Comorbidities and concerns of patients are comparable to diagnoses addressed in other adult neurology clinics. More research is needed to explore the impact of genetic testing on future care goals.

Figure 1: Reason for Visit

- Developmental delay
- Intellectual disability
- Autism
- Asperger’s syndrome
- Cerebral palsy
- Anxiety
- Epilepsy
- Motor speech disorder
- Sleep apnea
- Serum vitamin levels

Figure 2: Reason for Visit

- Developmental delay
- Intellectual disability
- Autism
- Epilepsy
- Motor speech disorder
- Sleep apnea
- Serum vitamin levels

Table 1: Demographics of Patients

<table>
<thead>
<tr>
<th>Sex assigned at birth</th>
<th>Gender identity</th>
<th>Race</th>
<th>Age distribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female</td>
<td>Female</td>
<td>White</td>
<td>25-30%</td>
</tr>
<tr>
<td>Male</td>
<td>Male</td>
<td>Black</td>
<td>10-15%</td>
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</tbody>
</table>

Table 2: Yield of Genetic Testing

<table>
<thead>
<tr>
<th>Genetic Test Ordered in Clinics</th>
<th>Total Ordered</th>
<th>Pathogenic results</th>
<th>Negative results</th>
<th>Variant of Uncertain Significance</th>
<th>Yield (% pathogenic)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chromosomal Microarray</td>
<td>11</td>
<td>3</td>
<td>6</td>
<td>2</td>
<td>27%</td>
</tr>
<tr>
<td>Fragile X Panel</td>
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<td>0</td>
<td>0</td>
<td>0</td>
<td>0%</td>
</tr>
<tr>
<td>Autism Spectrum Disorder/Intellectual Disability Panel</td>
<td>10</td>
<td>3</td>
<td>3</td>
<td>0</td>
<td>30%</td>
</tr>
<tr>
<td>karyotyping</td>
<td>4</td>
<td>0</td>
<td>4</td>
<td>0</td>
<td>0%</td>
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<tr>
<td>whole exome sequencing</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0%</td>
</tr>
</tbody>
</table>

Figure 3: Genetic Diagnoses Identified

- Trisomy 21
- Fragile X Syndrome
- Deletion syndromes
- Microduplications
- Cystic fibrosis
- Cerebral palsy
- Intellectual disability

Acknowledgements

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