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).[1] 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.[2,3] The lack of coordination and paucity of skilled services in all life stages can lead to significant health disparities.[4] 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 (in 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)/Intellectual 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.

Table 1: Demographics of Patients

Table 1
Demographics of Patients seen in the NDD Clinic

Baseline Characteristics (n = 86)	n (%)
Age at First Office Visit	
Less than 20 years	9 (11)
20-29 years	40 (47)
30-39 years	11 (13)
40-49 years	9 (11)
50-59 years	12 (14)
Over 60 years	5 (6)
Sex Assigned at Birth	
Male	59 (69)
Female	25 (29)
Unknown or not reported	2 (2)
Gender Identity	
Male	56 (65)
Female	27 (31)
Non-Binary	2 (2)
Unknown or not reported	1 (1)
Race	
White	66 (77)
Other	7 (8)
Asian	5 (6)
Black or African American	4 (5)
More than one race	4 (5)
Ethnicity	
Not Hispanic or Latino	69 (80)
Hispanic or Latino	16 (19)
Unknown or not reported	1 (1)
Home Location	
Urban	80 (93)
Rural	6 (7)
Living Situation	
Lives with parent(s) or Other Family Member(s)	57 (66)
Lives independently	14 (16)
Lives in a Group or Host Home	11 (13)
Lives in Supportive Independent Living	2 (2)
Lives in a Facility	2 (2)
Primary Caretaker	
Parent or Other Family Member	44 (51)
Patient by themself	27 (31)
Group or Host Home Caretaker	12 (14)
Facility Provider	2 (2)
Supportive independent living caretaker	1 (1)
Guardianship Status	
Patient has a guardian	40 (47)
Patient is their own guardian	35 (41)
Patient has a power of attorney (POA)	11
Patient has a healthcare proxy (HCP)	5
Patient has both POA and HCP	1
Patient has neither POA or HCP	18
Unknown or Not Reported	7 (8)
Patient is in the process of getting a guardian	4 (5)
Insurance Types	
Medicaid	40 (47)
Medicaid and Medicare	22 (26)
Private	12 (14)
Private and Medicaid	7 (8)
Medicare	4 (5)
Private and Medicare	1 (1)
BMI at First Office Visit	
Underweight (<18.5)	2 (3)
Healthy Weight (18.5-25)	27 (44)
Overweight (25-30)	16 (26)
Obese (>30)	14 (23)
Unknown or not reported	2 (2)
Not applicable (Telehealth visit)	25 (29)

Figure 1: Reason for Visit

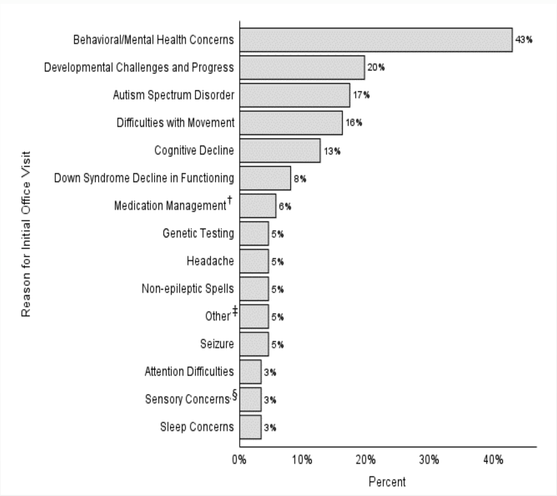

Figure 2: Reason for Visit

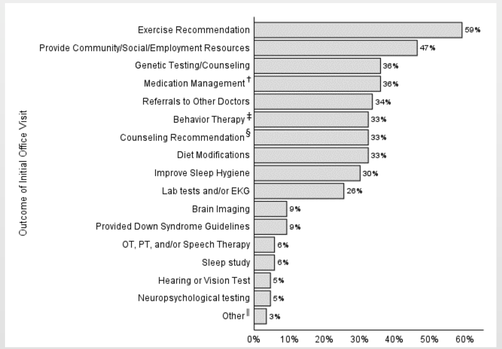

Table 2: Yield of Genetic Testing

Genetic Test Ordered in Clinics	Total Ordered	Pathogenic results	Negative Results	Variant of Uncertain Significance	Yield (% pathogenic)
Chromosomal Microarray	11	3	6	2	27%
Fragile X Panel	8	0	8	0	0%
Autism Spectrum Disorder/Intellectual Disability Gene Panel	10	3	3	4	30%
Exome Sequencing	4	2	1	1	50%
Genome Sequencing	1	0	0	1	0%

Figure 3: Genetic Diagnoses Identified

Sex assigned at birth	Previous Testing	Family history of ID/ASD	Diagnostic Test	Diagnosis	Common Features
Male	Fragile X	No	CMA	2p23.3 deletion (mosaic)	ID, distinct facial features, hypotonia
Female	Fragile X	Yes	CMA	16p11.2 duplication syndrome	DD/ID, ASD, psychiatric conditions
Male	Fragile X	Yes	CMA	22q11.2 deletion syndrome; Becker muscular dystrophy (incidental)	DD, ASD, distinct facial features, psychiatric conditions, congenital heart disease, palatal abnormalities; progressive muscle weakness
Male	None	No	ASD/ID panel	Witteveen-Kolk syndrome (SIN3A)	ID, distinct facial features, ASD
Male	None	No	ASD/ID panel	1q41-q42.12 deletion syndrome	ID, distinct facial features, seizures, midline defects
Female	CMA	No	WES	HNRNPU-related disorder	ID, early-onset epilepsy, ASD
Female	Fragile X, CMA	No	WES	Coffin-Siris syndrome (ARID1A)	ID, distinct facial features, hypoplasia of 5th digit
Male	None	No	ASD/ID panel	Phelan-McDermid syndrome (22q13.31q13.33 deletion)	ID, absent or delayed speech, distinct facial features
Male	None	No	WGS	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies	ID, global DD, absent or hypoplastic corpus callosum, distinct facial features, neonatal hypotonia
Female	Fragile X	Yes	ASD/ID Panel	Trisomy X	ASD, DD, learning disabilities, ADHD, anxiety, tall stature, hypotonia, premature ovarian insufficiency, renal abnormalities, seizures

CMA. Chromosomal Microarray; Fragile X. Fragile X Syndrome; ASD. Autism Spectrum Disorder; ID. Intellectual Disability; WES. Whole Exome Sequencing; DD. Developmental Disability

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 of specialized adult neurodevelopmental clinics. Comorbidities and concerns of patients seen 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.

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