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Research Letter | Genetics and Genomics

Genetic Testing Among Medicaid-Insured Children With Autism and Intellectual Disability

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Introduction

Autism spectrum disorder (ASD) and intellectual disability (ID) are pediatric conditions that benefit from early diagnosis and treatment,[1] and professional society guidelines recommend etiological genetic testing upon diagnosis.[2,3] A genetic diagnosis can enhance medical surveillance and lead to actionable therapies and specialized care.[2,4] Understanding factors affecting access to genetic testing is essential to ensure these benefits for individuals with ASD and/or ID. This cohort study examines testing frequency among children with ASD and/or ID across demographic factors using Medicaid claims data.

Methods

This cohort study included participants aged 7 to 17 years in 2019 who were enrolled in Medicaid each year from January 1, 2017, to December 31, 2019. Diagnoses were classified based on established algorithms using the *International Classification of Diseases, Ninth Revision* and *Tenth Revision* diagnostic codes,[5] which were compared with a random sample of children without ASD and/or ID and participants with epilepsy-only diagnostic codes (eMethods and eTable 1 in Supplement 1). Genetic testing was categorized using Current Procedural Terminology Codes (eTable 2 in Supplement 1). Logistic regression models estimated odds ratios (ORs) and 95% CIs, adjusting for covariates using SAS version 9.4 (SAS Institute). The institutional review board of Drexel University granted a HIPAA waiver. We followed the STROBE reporting guideline. Consent was not required because this was a secondary analysis.

Results

Our cohort included 885 895 children (mean [SD], 11.82 [3.07] years; 584 327 males [65.96%] and 301 568 females [34.04%]): 388 715 with ASD only, 122 792 with ID only, 82 107 with ASD and ID, 3306 with epilepsy only, and 292 281 without ASD or ID diagnosis codes (**Table 1**). Receipt of genetic testing during the 3-year study period was 8.44% for children with ASD and ID, followed by 4.80% for ID only and 4.17% for ASD only. The prevalence of genetic testing was lower among children diagnosed with epilepsy (2.44%) and still lower for the general pediatric sample (0.39%). Adjusted odds ratios (AOR) of genetic testing were 10.59 (95% CI, 10.22-11.54) for ASD only, 12.72 (95% CI, 11.94-13.56) for ID only, and 23.17 (95% CI, 21.75-24.69) compared with the random sample. AORs varied to a smaller extent by demographic and geographic factors (Table 1). Females had higher odds than males, and most racial and ethnic groups had lower adjusted odds of genetic testing compared with White children. Children residing in large rural towns had slightly higher odds of genetic testing.

From 2017 to 2019, Medicaid genetic testing claims saw declines in cytogenetics (21.5% to 18.8%), fragile X (16.8% to 15.0%), and microarrays (18.3% to 16.9%). In contrast, gene panel and exome sequencing shares increased from 41.8% to 46.6% and 1.6% to 3.0%, respectively (**Table 2**).

+ Supplemental content

Author affiliations and article information are listed at the end of this article.

Discussion

During the study period, we found that over 90% of eligible children with ASD only, ID only, and ASD and ID didn't receive genetic testing. There was only a modestly higher prevalence of testing in individuals with ASD and/or ID compared with individuals with epilepsy only and in the general population. Children with ASD and ID had the highest prevalence of testing, followed by ID only and ASD only. Females and those in large rural cities were more likely to be tested. Most racially and ethnically minoritized groups were less likely to undergo testing compared with White children. Gene

Table 1. Patient Characteristics and Adjusted Odds Ratios (aORs) of Genetic Testing

Characteristics	All, AOR (95% CI)[a,b]	ASD only		ID only		ASD and ID		Patient, No. (%)	
		No. (%)	AOR (95% CI)[c]	No. (%)	AOR (95% CI)[c]	No. (%)	AOR (95% CI)[c]	Random Sample	Epilepsy-only
All (N = 885 895)		388 715 (43.88)	NA	122 792 (13.86)	NA	82 107 (9.27)	NA	292 281 (32.99)	3306 (0.37)
Genetic Testing	NA	16 211 (4.17)	NA	5891 (4.80)	NA	6931 (8.44)	NA	1151 (0.39)	74 (2.24)
Age in 2019, mean (SD)	NA	11.52 (3.05)	NA	12.49 (2.96)	NA	12.58 (3.00)	NA	11.71 (3.08)	11.87 (3.07)
Sex									
Male	1 [Reference]	306 057 (78.74)	1 [Reference]	71 413 (58.16)	1 [Reference]	60 194 (73.31)	1 [Reference]	146 663 (50.18)	1745 (52.78)
Female	1.07 (1.04-1.09)	82 658 (21.26)	1.03 (0.99-1.07)	51 379 (41.84)	1.09 (1.04-1.15)	21 913 (26.69)	1.102 (1.04-1.16)	145 618 (49.82)	1561 (47.22)
Race									
American Indian or Alaska Native	0.69 (0.61-0.78)	4182 (1.08)	0.68 (0.56-0.80)	1286 (1.05)	0.74 (0.56-0.98)	761 (0.93)	0.8 (0.61-1.05)	4227 (1.45)	47 (1.42)
Asian or Pacific Islander	0.66 (0.63-0.68)	9627 (2.48)	0.71 (0.68-0.75)	4130 (3.36)	0.55 (0.51-0.60)	2918 (3.55)	0.68 (0.63-0.73)	12 110 (4.14)	86 (2.60)
Black or African American	0.90 (0.84-0.97)	62 711 (16.13)	1.02 (0.93-1.13)	28 989 (23.61)	0.72 (0.61-0.84)	15 837 (19.29)	0.96 (0.84-1.10)	61 796 (21.14)	784 (23.71)
Hispanic or Latino	0.88 (0.86-0.91)	99 354 (25.56)	0.90 (0.86-0.94)	28 289 (23.04)	0.96 (0.90-1.03)	19 362 (23.58)	0.88 (0.82-0.94)	101 645 (34.78)	959 (29.01)
Multiracial	0.82 (0.72-0.92)	4205 (1.08)	0.85 (0.73-1.00)	883 (0.72)	0.68 (0.48-0.96)	967 (1.18)	0.82 (0.65-1.04)	1562 (0.53)	31 (0.94)
White	1 [Reference]	165 007 (42.45)	1 [Reference]	42 061 (34.25)	1 [Reference]	33 369 (40.64)	1 [Reference]	93 246 (31.90)	1231 (37.24)
Missing or unknown	0.99 (0.95-1.03)	43 629 (11.22)	1.02 (0.97-1.08)	17 154 (13.97)	0.93 (0.86-1.01)	8893 (10.83)	1.018 (0.94-1.10)	17 695 (6.05)	168 (5.08)
Urbanicity									
Urban	1 [Reference]	324 347 (83.44)	1 [Reference]	98 744 (80.42)	1 [Reference]	67 877 (82.67)	1 [Reference]	244 893 (83.79)	2742 (82.94)
Large rural city/town	1.05 (1.01-1.09)	35 782 (9.21)	1.06 (1.01-1.12)	13 886 (11.31)	0.98 (0.90-1.06)	8191 (9.98)	1.09 (1.00-1.18)	26 394 (9.03)	313 (9.47)
Small rural town	1.04 (0.98-1.09)	16 960 (4.36)	0.99 (0.90-1.09)	6308 (5.14)	0.94 (0.84-1.06)	3693 (4.50)	1.10 (0.98-1.23)	12 586 (4.31)	143 (4.33)
Isolated small rural town	0.99 (0.92-1.06)	10 382 (2.67)	0.99 (0.90-1.09)	3418 (2.78)	0.95 (0.81-1.11)	2100 (2.56)	1.01 (0.87-1.18)	7843 (2.68)	NR[d]
Missing or unknown	0.87 (0.70-1.08)	1244 (0.32)	0.86 (0.64-1.16)	436 (0.36)	0.77 (0.47-1.24)	246 (0.30)	0.93 (0.59-1.48)	565 (0.19)	NR[d]

Abbreviations: AOR, adjusted odds ratio; ASD, Autism spectrum disorder; ID, intellectual disability; NR, not reported; NA, not applicable,

[a] Model adjusted for diagnosis group, sex, race and ethnicity, and urbanicity.

[b] Includes all children with ASD only, ID only, ASD and ID, and random sample.

[c] Model adjusted for sex, race and ethnicity, and urbanicity.

[d] Not reported due to the requirements of the Centers of Medicare & Medicaid Services.

Table 2. Genetic Testing Types by Study Year—All ASD and/or ID Children[a]

Year	Patients, No. (%)				
	Cytogenetics	Fragile X	Microarray	Gene Panel	Exome or genome
2017	7261 (21.53)	5656 (16.77)	6157 (18.26)	14 091 (41.79)	548 (1.63)
2018	5542 (20.39)	4406 (16.21)	4859 (17.87)	11 681 (42.97)	698 (2.57)
2019	4895 (18.83)	3907 (15.03)	4386 (16.88)	12 104 (46.57)	787 (3.03)

Abbreviations: ASD, Autism spectrum disorder; ID, intellectual disability.

[a] One child may have more than 1 genetic testing record.

panels were the most common genetic test, with slight growth from 2017 to 2019, replacing cytogenetics, fragile X, and microarrays. Exome sequencing nearly doubled but remained low.

These findings revealed gaps in genetic testing use among older Medicaid-insured children and adolescents with ASD or ID and highlight demographic and geographic differences. This corresponded with earlier findings of barriers to clinical genetic testing in specific subpopulations and suggested that clinical practices continue to not align with professional guideline recommendations.[6]

This study has limitations. Medicaid claims data have limited generalizability, lack clinical details, which hinders the establishment of causality between diagnoses and genetic testing, and have gaps in reporting for race and ethnicity. The sample restriction to children age 7 years and older, which was selected because many diagnoses of ID are made after age 6 years, excludes genetic testing that occurred before age 5 years, which may underestimate the prevalence of genetic testing. Our study data are limited to the 2017 through 2019 period, but they provide the most up-to-date reporting available without confounding access barriers driven by the COVID-19 pandemic. Future research should explore health policies and interventions to improve access to genetic testing services among individuals with ASD or ID.

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SUPPLEMENT 1.

eMethods.

eTable 1. *International Classification of Diseases, Ninth Revision* (*ICD-9*) and *Tenth Revision* (*ICD-10*) diagnosis codes

eTable 2. Genetic testing CPT codes

eReferences.

SUPPLEMENT 2.

Data Sharing Statement

Supplemental Online Content

Brown TR, Lee WL, Ventimiglia J, et al. Genetic testing among Medicaid-Insured children with autism and intellectual disability. *JAMA Netw Open*. 2025;8(9):e2533518. doi:10.1001/jamanetworkopen.2025.33518

This supplemental material has been provided by the authors to give readers additional information about their work.

eMethods

We utilized longitudinal administrative data from the Transformed Medicaid Statistical Information System (T-MSIS) Analytic Files (TAF) database, covering the period from January 1, 2017, to December 31, 2019, from the Centers for Medicare and Medicaid Services (CMS). The analysis was completed from June 18, 2024, to June 27, 2024. This dataset includes information on enrollment, diagnosis codes, inpatient services, outpatient services, and outpatient drug claims. Individual demographic details are contained in personal summary files. Individuals without information on race, ethnicity, or urbanicity were classified as missing. Race and ethnicity were optionally reported by Medicaid and CHIP beneficiaries (i.e., parent reports) and collected at the state level using a variety of classifications differing in the names and numbers of categories as well as how those categories were combined to create aggregated race/ethnicity categories that conformed to OMB guidelines. States reported to CMS using their own classifications, which were recategorized by CMS under the following combined race and ethnicity categories[1]: American Indian and Alaska Native; Asian, Black, Hawaiian/Pacific Islander; Hispanic; Multiracial; White; or other, which includes non-Hispanic with missing race and missing both race and ethnicity. The race and ethnicity represent the original database item. The study population consisted of children aged 7–17 in 2019 who had complete data throughout the study period and were enrolled in Medicaid for at least 9 months each calendar year from 2017 to 2019. Children with any third-party liability insurance coverage, including private insurance coverage outside of Medicaid, whether primary or secondary, during the study period were excluded. “ASD-only“ refers to having at least one inpatient claim or two non-drug claims of any service type linked to diagnostic codes for ASD (eTable 1), without the presence of co-occurring ID. “ID-only“ refers to having at least one inpatient claim or two non-drug claims of any service type linked to diagnostic codes for ID (eTable 1), without the presence of co-occurring ASD. “Epilepsy-only”, refers to the presence of Epilepsy diagnostic codes without the presence of co-occurring ASD or ID. Diagnoses are based on established algorithms from the Chronic Conditions Data Warehouse[2]. This study includes a randomly selected comparison cohort of Medicaid-enrolled children without ASD or ID ICD-9/10 diagnosis codes who met the inclusion criteria of 9 months of enrollment each study year. Epilepsy-only was included because it is not a neurodevelopmental diagnosis, and it has its own guidelines for genetic testing to act as a comparison to

the genetic testing frequency in ASD and ID. The primary outcome was genetic testing evaluated the presence of Current Procedural Terminology (CPT) Codes (eTable 2). No adjustments were made for multiple comparisons. Additional details regarding study methods are available as previously published[3].

eTable 1. ***International Classification of Diseases, Ninth Revision*** **(*ICD-9*) and** ***Tenth Revision*** **(*ICD-10*) Diagnosis Codes**

Diagnosis	Reference Period (years)	ICD-9 Codes	ICD-10 Codes	Type of Algorithm	Years of data used	Continuous enrollment requirements (months)[a]	Age range in years in 2016
Autism Spectrum Disorders	2	299.0, 299.00, 299.01, 299.1, 299.11, 299.8, 299.80, 299.81, 299.9, 299.90, 299.91	F84.0, F84.3, F84.5, F84.8, F84.9	At least 1 inpatient claim OR 2 other non-drug claims of any service type with DX codes (any diagnosis on the claim)	2017 to 2019	9	7 - 17
Intellectual Disabilities and Related Conditions	2	317, 318, 318.0, 318.1, 318.2, 319,	F70, F71, F72, F73, F78, F79	At least 1 inpatient claim OR 2 other non-drug claims of any service type with DX codes (any diagnosis on the claim)	2017 to 2019	9	7 - 17
Epilepsy	2	DX 345, 345.0, 345.00, 345.01, 345.1, 345.10, 345.11, 345.2, 345.3, 345.4, 345.40, 345.41, 345.5, 345.50, 345.51, 345.6, 345.60, 345.61, 345.7, 345.70, 345.71, 345.8, 345.80, 345.81, 345.9, 345.90, 345.91	DX G40.001, G40.009, G40.011, G40.019, G40.101, G40.109, G40.111, G40.119, G40.201, G40.209, G40.211, G40.219, G40.301, G40.309, G40.311, G40.319, G40.401, G40.409, G40.411, G40.419, G40.42, G40.501, G40.509, G40.801, G40.802, G40.803, G40.804, G40.811, G40.812, G40.813, G40.814, G40.821, G40.822, G40.823, G40.824, G40.833, G40.834, G40.89, G40.901, G40.909, G40.911, G40.919, G40.A01, G40.A09, G40.A11, G40.A19, G40.B01, G40.B09, G40.B11, G40.B19, G40.C01, G40.C09, G40.C11, G40.C19	At least 1 inpatient claim OR 2 other non-drug claims of any service type with DX codes (any diagnosis on the claim)	2017 to 2019	9	7 - 17

[a] 9-month out of 12-month enrollment was required to account for continuity of enrollment by small disruptions of Medicaid enrollment due to administrative processes[4].

eTable 2. Genetic Testing CPT Codes

CPT Code	Domain
81228;81229	Arrays
81245;81246;81247;81415;81416;81417;81425;81426;81427	Exome / Genome Sequencing
81243;81244;83897;83900;83909;83891	Fragile X
88271;88272;88273;88274;88275	Molecular cytogenetics
83890;83891;83892;83893;83894;83896;83898;83901;83902;83903;83904;83905;83906;83907;83908;83912;83913;83890;83892;83893	Other
81185;81186;81239;81302;81303;81304;81321;81322;81323;81331;81400;81401;81402;81403;81404;81405;81406;81407;81408;81419;81442;81470;81471;81479	Single genes / Panels
88230;88245;88248;88249;88260;88261;88262;88263;88264;88267;88269;88280;88283;88285;88289;88291	Traditional cytogenetics

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Data Sharing Statement

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Data
Data available: No