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Short Report: Prevalence of Autism Spectrum Disorder in a Large Pediatric Primary Care Network

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Lay Abstract:

Historically, children from non-Hispanic Black and Hispanic backgrounds, those from lower-income families, and girls are less likely to be diagnosed with autism spectrum disorder (ASD). Under-identification among these historically and contemporaneously marginalized groups can limit their access to early, ASD-specific interventions, which can have long-term negative impacts. Recent data suggest that some of these trends may be narrowing, or even reversing. Using electronic health record data, we calculated ASD prevalence rates and age of first documented diagnosis across socio-demographic groups. Our cohort included children seen at young ages (when eligible for screening in early childhood) and again at least after age 4 years in a large primary care network. We found that ASD prevalence was unexpectedly higher among Asian children, non-Hispanic Black children, children with higher Social Vulnerability Index scores (a measure of socio-economic risk at the neighborhood level), and children who received care in urban primary care sites. We did not find differences in the age at which ASD diagnoses were documented in children's records across these groups. Receiving primary care at an urban site (regardless of location of specialty care) appeared to account for most other socio-demographic differences in ASD prevalence rates, except among Asian children, who remained more likely to be diagnosed with ASD after controlling for other factors. We must continue to better understand the process by which children with ASD from traditionally under-identified and under-served backgrounds come to be recognized, to continue to improve the equity of care.

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Background:

Socio-demographic inequities in autism spectrum disorder (ASD) diagnosis have been documented for two decades (Mandell, Listerud, Levy, & Pinto-Martin, 2002). Missed diagnoses can result in delayed or no access to ASD-specific services, with negative long-term consequences. Most previous studies, including large cohort studies with rigorous case definitions, have found that Non-Hispanic White children (hereafter referred to as "White") are diagnosed at higher rates and earlier ages than Non-Hispanic Black (hereafter referred to as "Black") and Hispanic children, and that children from families with higher socio-economic status (SES) are diagnosed at higher rates and earlier ages than children from lower-SES families (Baio et al., 2018).

Race and ethnicity are socio-political constructs. Thus, differences in rates or ages of diagnoses are more likely due to differences in service systems (Tregnago & Cheak-Zamora, 2012), biases (Broder-Fingert, Mateo, & Zuckerman, 2020), and imperfect screening and diagnostic tools (Guthrie et al., 2019). Examining prevalence differences within a system where children theoretically have more similar access to screening and diagnostics can help elucidate the pathways for inequities that may exist.

Recent data paints a different picture of diagnoses. Surveillance data among 4 year-old children in the U.S. from the Autism and Developmental Disabilities Monitoring (ADDM) Network found that White children had lower prevalence than Black, Hispanic, and Asian/Pacific Islander children; children with the lowest median household income had a higher prevalence than children with the highest (Shaw KA, 2021). Survey data using parent-reported ASD diagnoses found a prevalence rate of 3.14, with no statistically significant differences by race or ethnicity (Li et al., 2022). And a recent cohort study in England found higher prevalence among Black students than any other racial or ethnic group, and among children who received free meals at school (Roman-Urrestarazu et al., 2021). Similarly, a recent large population-based study found that the prevalence of ASD was higher among publicly versus privately insured children (Straub et al., 2022).

Using data from an integrated primary care and subspecialty network, we examined medical records of children seen in primary care at eligible ASD screening ages and followed through at least 4 years of age to examine the prevalence of ASD; age of first documented ASD diagnosis; and whether the prevalence and age of documented diagnosis varied by race, ethnicity and SES.

Methods:

This study retrospectively analyzed a cohort of children who received primary healthcare within the Children's Hospital of Philadelphia (CHOP) Care Network, a large pediatric health system that provides primary and subspecialty services, including ASD diagnostics by developmental-behavioral pediatricians, neurologists, psychiatrists or psychologists. The Care Network includes 29 pediatric primary care sites in PA and NJ, which have conducted universal ASD screening since 2014 and have achieved high screening rates, with 91% of

eligible children screened at least once (Guthrie et al., 2019). Children identified as at risk can be internally referred for ASD assessment or can access evaluations in the community.

This same cohort was previously used to assess the accuracy of universal autism screening (Guthrie et al., 2019) and includes 23,015 children who presented at least once for a well-child visit between 16 and 26 months of age at a CHOP primary care site between 2011 and 2015 and again after age 4 (to assess diagnostic outcome). The present sample includes additional diagnostic outcome data available after our previous study was published in 2019. Children for this study were median 9.51 years-old (interquartile range, IQR 8.97–10.42 years) when data were extracted.

A child was considered to have ASD if this diagnosis appeared in the electronic health record (EHR) more than once (in the problem list or as a visit diagnosis) or was provided by an ASD specialist (Guthrie et al., 2019). The date at which the ASD diagnosis first appeared in the EHR was used as a proxy measure for age of diagnosis. A diagnosis could appear because a child was diagnosed by a CHOP subspecialist in developmental and behavioral pediatrics, neonatal follow-up, neurology, psychiatry, and/or psychology (62.6% of diagnoses), or because a diagnosis was recorded twice by a CHOP clinician (37.4% of diagnoses). All available data at the time of electronic data extraction (December 2021) were used to determine diagnostic outcome (Guthrie et al., 2019). Manual review of 10% of charts with a first ASD diagnosis recorded after our original data extraction confirmed accuracy of newly documented diagnoses.

Socio-demographic variables were digitally extracted from the EHR and categorized to identify possible disparities in care: sex (*Male/Female*), parent-reported race and ethnicity (*Asian; Black; Hispanic; White; and Multiple/Other races*), preferred language (*English, Spanish, or Other language*), insurance type *(Public/Medicaid or Private*), site of care at the screening visit (*urban, suburban/rural*), and Social Vulnerability Index (SVI) at the census tract level. The SVI uses percentile rankings for 15 census variables from the *American Community Survey* to compute an overall vulnerability score for each census tract compared to others, based on composite measures of SES, household composition and disability, minority status and language, and housing and transportation (Flanagan, Hallisey, Adams, & Lavery, 2018). The SVI is based on patient addresses at the time of screening, with higher percentages indicating higher levels of social vulnerability or relative disadvantage; we present the overall SVI percentage and nationally defined mutually exclusive quartiles (*High, Medium high, Medium low, and Low)*.

Statistical Analyses:

ASD prevalence rates and age of documented diagnosis were compared for race and ethnicity, SVI, and site of care using chi-square tests, t-tests and ANOVA as appropriate, with pairwise comparisons for multi-category predictor variables. Reference groups were selected based on groups that historically had highest rates of ASD diagnosis. Socio-demographic variables with statistically significant univariate associations with ASD diagnosis were included in hierarchical regression models in a step-wise fashion to examine the additive effects of these variables on outcomes of interest. SVI was analyzed separately as a continuous and a categorical variable, with similar results. We did not include insurance

type in regression models, as this socio-demographic measure is redundant with SVI, or sex, as sex-based ASD differences are well documented (Shaw KA, 2021). Analyses were conducted in SPSS version 26 (*IBM Corp*, 2019); assumptions for all parametric tests (which are robust to moderate non-normality) were met. This protocol was approved by CHOP's Institutional Review Board. There was no community involvement in this project.

Results:

Across the entire cohort (N= 23,015), 41.1% identified as White, 36.5% as Black, 11.2% as another race or multiple races, 7.3% as Hispanic, and 3.9% as Asian; 45.4% were publicly insured (*see* Table 1). These demographics generally reflect the racial composition of Philadelphia and the surrounding region, although Asian children were under-represented as were uninsured children ("Data USA: Philadelphia, PA,"). The prevalence of ASD was 3.2% across the entire cohort, with a median age of diagnosis of 3.93 years. The following groups had higher ASD prevalence (Table 2): Asian children (5.4; odds ratio, OR=2.5, p<.001) and Black children (3.5%, odds ratio, OR=1.3, p=.002) compared to White children (2.7%); publicly (3.6%; OR=1.28, p<.001) versus privately insured children (2.8%); those with highest quartile SVI (3.6%, OR=1.47, p<.001), Medium High (3.4%, OR=1.38, p<.003), and Medium Low 3.4%, (p=1.37, p=.006) compared to Lowest quartile SVI (2.5%); and children seen in urban (3.8%, OR=1.38, p<0.001) versus suburban/rural primary care sites (2.8%). No statistically significant differences in age of ASD documentation in the EHR were identified for any of the socio-demographic factors, and as such, multivariable models were not run.

Hierarchical logistic regressions were conducted to identify the individual effects of the socio-demographic variables on ASD rates. Race/ethnicity was added to the model in the first step, demonstrating differences in ASD rates for Asian and Black compared to White children. This difference for Asian children remained significant (p<.001) when SVI (p<.02)was added to the model in the second step, but the difference for Black children did not. When site (aOR=1.25, p<.04) was added to the model in the third step, the difference for Asian children remained significant (aOR=1.82, p<.001), but the effect of SVI did not.

Discussion:

Across this cohort, ASD prevalence (3.2%) was higher than in previously described samples, but similar to recent estimates from 2019–2020 (3.14%) (Li et al., 2022). The median age of first diagnosis was just under 4 years.

These analyses were exploratory in nature and meant to be hypothesis-generating. We posit several explanations for our high overall prevalence rate, noting some that may be unique to our healthcare system. First, our prevalence rate might be accurate and timely, matching data from 2019–2020 that found an estimated prevalence of 2.79–3.49% (Li et al., 2022). Alternatively, families of children with ASD might be more likely to continue to seek primary care at an academic children's network compared to community sites (e.g., selection bias), and thus may be over-represented in a cohort followed by CHOP primary care. Another alternative is that our high prevalence rate might reflect diagnostic substitution,

as the linking of effective ASD therapies may encourage diagnosis of ASD rather than language or developmental disorders alone, or the imprecision that can be associated with ASD diagnosis (Mottron, 2021). Lastly, we represent lifetime prevalence, while the possibility exists that some children have documented diagnoses that later are removed as a result of earlier inaccuracy or changes in core features. Each of these potential explanations warrants additional study to determine what clinical-, individual- or systems-level factors contribute to ASD prevalence and inequities therein.

Contrary to our hypothesis, ASD prevalence differed by socio-demographic variables. Though effect sizes were generally small (Chen, Cohen, & Chen, 2010), there was an even higher prevalence among minoritized children than recent trends would suggest. Socio-demographic differences in median age of diagnosis were not found. The rate among Asian children in this cohort was particularly high (5.4%).

Our data came from children seen for primary care at urban and suburban practices across several counties with different services. Being seen at an urban (Philadelphia) primary care site was an important predictor of ASD diagnosis, and appeared to account for most other socio-demographic differences. The urban sites have closer proximity to the academic medical center and more academic clinicians and trainees who might have more training in ASD recognition. Children in Philadelphia live closer to specialty diagnostic care, either at CHOP or in the community. Finally, publicly funded Early Intervention services in Philadelphia are strong, including for ASD, which may increase attention to the need for diagnosis to access these services.

There are multiple points along the screening, diagnostic and therapeutic process that may contribute to diagnostic differences (Wallis, 2021). These findings suggest that systemic factors (such as geographic access to a diagnostic clinic) play a large role in determining diagnosis. More work is needed to better understand the complexity of these processes, and the ways that systems may contribute to inequities (Broder-Fingert et al., 2020).

There are several limitations to the current study. We used a retrospective clinical cohort. Thus, we were unable to confirm the accuracy of ASD diagnoses. However, the stringent criteria used for ASD diagnosis has previously been validated as highly accurate in other large health systems using EHR/claims data (Coleman et al., 2015), and our manual chart review provided sufficient documentation to justify diagnosis for the majority of children. However, some children might have been missed and some inappropriately classified as having ASD. Children in our cohort had varying lengths of follow-up (i.e., 4–11 years), such that some younger children may not have been diagnosed yet, which may have downwardly biased age of first diagnosis estimates.

Additionally, there is heterogeneity among our racial and ethnic groups that could not be studied using EHR data (e.g., differences exist between and within Chinese and Indian cultures, but all might be classified as Asian). Important components of SES, such as parental education and maternal nativity are not available in the EHR. Furthermore, the finding of such a high prevalence rate in Asian children warrants further investigation, which we are examining separately. Findings may not generalize to other settings where

screening and diagnostic evaluations are not as easily accessible, or among different patient populations and clinical settings, or among populations that are un- or under-insured. We also did not examine service use; additional disparities in service access are possible and perhaps likely. Other sociocultural and historical factors (e.g., stigma, institutional racism, etc.) undoubtedly also play a role in ASD diagnosis and service utilization. The reasons behind our reported higher and varied diagnostic rates should be explored further.

Conclusion:

Systemic factors appear to contribute to socio-demographic differences in ASD diagnosis. Future work should aim to understand the ASD diagnostic process to ensure that the promise of early identification and intervention is realized for all children.

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Abbreviations:

ADDM	Autism and Developmental Disabilities Monitoring
ASD	Autism Spectrum Disorder (ASD)
CHOP	Children's Hospital of Philadelphia (CHOP)
EHR	Electronic health record (EHR)
SES	Socioeconomic status
SVI	Social Vulnerability Index

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Table 1:

Demographics of the total population, and cohort with ASD diagnosis

Characteristic	Total sample N= 23,015	ASD Diagnosis n= 731 Prevalence Rate= 3.2%
Age at screening visit in months *median* (IQR)	18.74 (18.18–20.52)	18.81 (18.2–21.3)
Age at most recent follow-up visit in years *median* (IQR)	8.85 (8.15–9.65)	8.98 (8.33–9.80)
Age of ASD Dx in years *Median (IQR)*	N/A	3.93 (2.83–6.08)
Chronologic age at time of data abstraction (December 2021) *Median (IQR)*	9.48 (8.96–10.45)	9.51 (8.97–10.42)
Sex *n* (%)		
Female	11,142 (48.4)	164 (22.4%)
Male	11,873 (51.6)	567 (77.6%)
Race/ethnicity *n* (%)		
Asian	888 (3.9)	48 (6.6)
Black, non-Hispanic	8,366 (36.5)	294 (40.2)
Hispanic	1,684 (7.3)	57 (7.8)
Other/Multiple Races	2,576 (11.2)	77 (10.5)
White, non-Hispanic	9,436 (41.1)	255 (34.9)
Family's Preferred Language *n* (%)		
English	20,043 (96.7)	678 (96.0)
Spanish	466 (2.2)	15 (2.1)
Other Language	225 (1.1)	13 (1.8)
Insurance Split *n* (%)		
Private	12,492 (54.6)	353 (48.7)
Public (Medicaid)	10,408 (45.4)	372 (51.3)
Site of Care at Screening Visit *n* (%)		
Urban	9613 (41.8)	362 (49.5)
Suburban/Rural	13,402 (58.2)	369 (50.5)
Social Vulnerability Index, *Mean (SD)*	0.49 (0.32)	0.53 (0.31)
Quartiles n (%)		
Highest	6585 (28.6)	238 (32.6)
Medium High	4813 (20.9)	164 (22.5)
Medium Low	4353 (18.9)	147 (20.1)
Lowest	7252 (31.5)	181 (24.8)
Diagnosis conferred by ASD subspecialist at CHOP only*, n (%)	N/A	25 (3.4)
Diagnosis with 2 mentions in EHR only	N/A	273 (37.4)
Both methods of diagnostic ascertainment	N/A	433 (59.2)

* Includes documentation of diagnosis by developmental and behavioral pediatrics, neonatal follow-up program, neurology, psychiatry, and/or psychiatry

Table 2:

Prevalence Rates and Comparisons by Socio-Demographic Group

Demographic Characteristic	**ASD Prevalence Rate n (%)**	**Odds Ratio of ASD diagnosis** (Chi Square, compared to reference group indicated)	**P value**	**Age of diagnosis** (ANOVA, not assuming equal variances)	**P value**
Race/ethnicity *n* (%)				*Mean (SD)*	
Asian	**48 (5.4)**	**2.06 (1.50–2.82)**	**<0.001**	4.27 (2.02)	
Black, non-Hispanic	**294 (3.5)**	**1.32 (1.11–1.56)**	**0.002**	4.73 (2.23)	
Hispanic	57 (3.4)	1.27 (0.94–1.69)	0.12	4.46 (2.13)	0.40
Other/Multiple Race	77 (3.0)	1.11 (0.86–1.44)	0.43	4.52 (2.19)	
White, non-Hispanic	255 (2.7)	Reference	Reference	4.45 (2.14)	
Insurance *n* (%)				*Mean (SD)*	
Public (Medicaid)	**372 (3.6)**	**1.28 (1.10–1.48)**	**0.001**	4.46 (2.10)	0.24
Private	**353 (2.8)**	**Reference**		4.65 (2.21)	
Social Vulnerability Index *(Continuous Percentile)*		t test, t statistic **−3.87**	t test **<0.001**	Pearson Correlation 0.01	t test 0.79
Social Vulnerability Index (*Quartiles)*				*Mean (SD)*	
Highest	**238 (3.6)**	**1.47 (1.20–1.78)**	**<0.001**	4.56 (2.20)	
Medium High	**164 (3.4)**	**1.38 (1.11–1.71)**	**0.003**	4.68 (2.24)	0.82
Medium Low	**147 (3.4)**	**1.37 (1.10–1.70)**	**0.006**	4.54 (2.06)	
Lowest	**181 (2.5)**	**Reference**	**Reference**	4.45 (2.09)	
Site of Care at Screening Visit *n* (%)					
Urban	**362 (3.8)**	**1.38 (1.19–1.60)**	**<0.001**	4.68 (2.26)	0.10
Suburban/Rural	**369 (2.8)**	**Reference**	**Reference**	4.43 (2.04)	

Regression Analyses Predicting ASD Diagnosis

Variables in the Equation	**Odds Ratio, Adjusted for Other Variables in the Equation (aOR)**	**P value**
Race/ethnicity *n* (%)		
Asian	**2.06 (1.50–2.82)**	**<0.001**
Black, non-Hispanic	**1.31 (1.10–1.55)**	**0.002**
Hispanic	1.26 (0.94–1.69)	0.12
Other/Multiple Race	1.11 (0.86–1.44)	0.43
White, non-Hispanic	Reference	Reference
Race/ethnicity *n* (%)		
Asian	**1.92 (1.40–2.65)**	**<.001**
Black, non-Hispanic	1.10 (0.88–1.37)	0.41
Hispanic	1.13 (0.83–1.53)	0.45
Other/Multiple Race	1.06 (0.81–1.38)	0.67

White, non-Hispanic	Reference	Reference
Social Vulnerability Index (Percentile)	**1.46 (1.07–1.98)**	**<0.02**
Race/ethnicity *n* (%)		
Asian	**1.82 (1.32–2.53)**	**<0.001**
Black, non-Hispanic	1.00 (0.79–1.28)	0.99
Hispanic	1.09 (0.80–1.49)	0.59
Other/Multiple Race	1.04 (0.80–1.36)	0.75
White, non-Hispanic	Reference	Reference
Social Vulnerability Index (Percentile)	1.26 (0.90–1.76)	0.19
Site of Care at Screening Visit *n* (%)		
Urban	**1.25 (1.01–1.54)**	**<0.04**
Suburban/Rural	Reference	Reference