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Duration of untreated autism in rural America: emerging public health crisis

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Abstract

The rural areas have been at the receiving end amidst mental health disparity across the USA. There is a serious and concerning divide among ones with autism spectrum disorders (ASDs) living in underserved areas as compared to urban residents. With the higher than ever prevalence of ASD as per the recent reports of the Centers for Disease Control and Prevention; there is a need for a closer look at the prevailing issues. The trends are reflecting marked underdiagnosis, late diagnosis, lack of evidence-based diagnostic measures and interventions. These factors interplay in worsening the mental health crisis and there is an urgent need for corrective measures to address these highly modifiable problems.

Introduction

Despite many advances in the knowledge of autism spectrum disorder (ASD), newer evidence is beyond the reach of the rural populations. The data of the mental health crisis reflects the significant disparity in quality, access, and costs. The increase in suicide rates among adolescents,[1] the opioid crisis in adults are a few recent trends that have bought widespread attention to the rural mental health crisis.[2]

ASD could be diagnosed at the age of 12 to 14 months,[3] and it is a highly stable diagnosis maintained at age 3.[4] It takes 36 months after parental concerns to have a formal diagnosis.[5-7] Parental concerns are known to detect 70% to 80% of children with disabilities.[8] The Autism and Developmental Disabilities Monitoring (ADDM) Network surveillance is critical to accurately measure epidemiology and has highlighted the variability in the prevalence. The average age of diagnosis is between 4 and 7 years,[9,10] and the global mean average age is around 60.8 months.[11]

In 2021, the Centers for Disease Control and Prevention (CDC) has changed the prevalence of ASD from 1 in 54 (1.9%) to 1 in 44 (2.3%) 8-year-old children.[12] There are many serious implications of this revised prevalence, first, the likelihood of underdiagnoses in children with specific ethnicity,[13] race,[14] and living in the underserved areas.[15] Second, if the prevalence was high then what happened to the undiagnosed, and finally, what is the extent of rural disparities.[16]

The 2016 U.S. Preventive Services Task Force recommendations against universal screenings were criticized[17] but given the newer data, is it a serious public health concern, it should be reconsidered? We appraise some critical determinants and their impact on the clinical practices in rural areas.

Duration of untreated autism

Late diagnosis of ASD is associated with a higher incidence of depression and self-harm in adolescents.[18] The ADDM reports the median age of ASD diagnosis among children under age 8 was lowest in the urban population of California, Maryland, and New Jersey with California having the lowest median age of 36 months. And longest in rural states of Minnesota (63 months), Arizona (58 months), and Wisconsin (56 months).[12] These data also suggest a significant disparity between states with the highest prevalence like California (1:26) with the lowest in Missouri (1:60).[12] The ADDM Network surveillance found the prevalence is similar among Caucasians, African Americans, and Hispanic nationally.[12] However, these data also vary between states with some states ASD is more prevalent among the Caucasian population while others like Maryland and Minnesota where ASD was more prevalent in an African American population.[12]

Females

The delay in females is more pervasive given only recently there is more recognition of a distinct phenotype[19] and also symptomatology is been different from males.[20] There have been recent efforts in modifying the Autism Diagnostic Observation Scale Second edition (ADOS-2) text to

identify females[21] but they continue to be underrepresented in the overall prevalence of ASD with a sex ratio of 4.2 in favor of males.[12]

Comorbidity

It is also been reported that ASD has many overlapping symptoms with ADHD.[22] It is widely accepted that the presence of comorbidity not only challenges treatment but also has diagnostic implications.[23,24] Another study suggests many with ASD were diagnosed with ADHD and as a result, there was a significant delay in the diagnosis.[22] Table 1.

The myths and utilities of testing

Diagnostic tools Autism Diagnostic Observation Scale Second edition, Autism Diagnostic Interview-Revised, Childhood Autism Rating Scale (ADOS-2, ADI, CARS) are recommended to be part of the multidisciplinary assessment in preschoolers[25] and not as stand-alone measures. Despite improved accuracy of the gold standard tools like ADOS-2[26], they may not identify all cases of ASD.[27] There are concerns about the cost-effectiveness of ADOS-2 and the need for extensive training requirements.[26] On comparative analysis of sensitivity, specificity, and the positive predictive value (PPV) of the diagnostic tools, ADOS was more sensitive but all three of them had the same specificity.[25]

Genetic testing

Chromosomal microarray analysis (CMA) has been recommended as the standard of care for the initial evaluation of children with developmental disabilities and/or ASDs.[28] The emergence of low-cost whole-exome sequence (WES) and whole-genome sequence (WGS), which are preferred over CMA.[29] A study has found 12% with ASD have pathologic findings on the genetic testing[30] and as per AACAP, it could be as high as one-third in suspected cases.[31] Genetic testing is rarely done in rural areas although there is no data to support it.

Reasons for late diagnosis in the rural US

Two recent studies found the sensitivity and PPV of the most commonly used screening tool the Modified Checklist for Autism

Table 1. The Summary of Facts Related to the Delay in the Diagnosis of Autism in Rural Areas

Late diagnosis of ASD
• In 2021, the prevalence of ASD as per CDC is changed to highest ever 1 in 44 (2.3%) for 8-year-old children
• ASD could be diagnosed at age 12 to 14 mo, but the average age of diagnosis is between 4 and 7 y
• Underdiagnoses are in children with specific ethnicities, races, and living in underserved areas
• The 2016 US Preventive Services Task Force recommendations against universal screenings
• The median age of diagnosis among children under age 8 was lowest in the urban areas of states and prevalence of highest in states with urban populations

Abbreviations: ASD, autism spectrum disorder; CDC, Centers for Disease Control and Prevention.

in Toddlers (MCHAT) was in the range of 33.1% to 38.8%, and 14.6% to 17.8%, respectively.[32,33] Both studies have called for a paradigm shift in screenings given current tools' has poor sensitivity and false negatives.[34,35]

A British population-based cohort found those living in poverty and with higher intelligence are likely to miss out on the initial screenings.[36] Therefore, testing preschoolers from low-income families and minorities may identify these children.[37] Multilingual staff may increase accuracy given MCHAT has a poor PPV in ethnic minority.[38] There is presence of a distinct subtle phenotype that lacks symptoms at early stages and therefore detection before age 4 is difficult due to the heterogeneous nature of ASD.[39] These studies emphasize the value of longitudinal follow-up beyond 36 months.[40]

There is a clear divide between the prevalence of ASD in states with urban populations (California and New Jersey) as compared to that with the rural population (Missouri and Wisconsin).[12] These differences in the prevalence could be attributed to the changes in the diagnostic criteria and higher health care literacy in urban areas.[41,42] The state public health awareness programs also contribute to improving awareness among parents.[43]

Evidence-based interventions which work is not available to many who needs the most

The higher prevalence could also be linked to the higher density of mental health workforce[44] with Maryland being the only outlier.[12] The services are clustered around urban pockets with higher per capita income. Also, the average cost for Applied Behavioral Analysis is $60,000 per year which totals to $240,000 for 4 years of treatment.[45]

Discussion

Currently, MCHAT is recommended at 16 months of age in suspected cases and universal screening is not recommended.[46] As the burden of missed or late diagnosis of ASD continues to mount, the scientific basis for controversial population-based universal screening growing. There is also a need for a screening tool that is highly specific, sensitive, and with a good PPV given MCHAT has both poor specificity and sensitivity.[47] Additionally, translated versions of MCHAT perform suboptimally.[48,49] Special attention is required for the underrepresented minority population. There is a need to modify questions in the ADOS-2 based upon the cultural context to reduce the diagnostics bias.[50] Even the Spanish-translated version is less accurate compared to English; despite Spanish being the second most spoken language in the US.[51] Cost-effective, readily accessible tools assist in early diagnosis and the access to interventions may not only improve the long-term outcomes but also reduce the overall cost. The multidisciplinary approaches for diagnostics and treatments are expensive to set up and continue to remain a barrier in underserved areas.[52,53]

The emerging evidence of artificial intelligence-based platforms is encouraging since it has the potential to address diagnostics issues and are also cost-effective.[54] Many diagnostic instruments that are underutilized, training staff to use ADI-R, ADOS-2, CARS-3, GADS may be highly prudent in rural areas with limited resources. The gold standard diagnostic tool ADOS is highly sensitive but lacks specificity.[25] This is observed in ones with fewer symptoms of ASD and when associated with comorbid ADHD or social anxiety disorder.[55] Therefore, in many with fewer symptoms

of ASD and associated comorbidities, diagnosis is more difficult.[23,24] The adults with high functioning ASD may compensate for their deficits, and never meet the criteria for autism even with ADOS-2.[56]

Finally, significant advocacy is needed for the rural low-income populations living in the underserved areas with a low incidence of ASD.

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