Differences in Autism Diagnosis Rates Due to Various Racial, Geographic, Genetic, and Biological Factors

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Abstract:

Autism spectrum disorder (autism) has core diagnostic features of differences in social behavior and restricted interests or repetitive behaviors. This paper provides a review of the global influence of race, geographic locations, and culture on autism diagnosis. Different geographic populations are known to share certain genetic patterns, and this review seeks to examine research about genetic patterns related to geographic location and its relation to autism. Racial norms, as well as their affiliated cultural beliefs, and language barriers can impact the identification and assessment of autism symptoms, leading to diagnostic disparities among diverse populations worldwide. Additionally, healthcare system factors, including resource availability and practitioner biases, can further contribute to variations in diagnostic outcomes. This review discusses the differences in autism diagnosis based on racial and geographic factors and suggests further actions and initiatives to increase equality in autism diagnoses.

Introduction:

Autism spectrum disorder (henceforth autism) is a neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction across multiple contexts, as well as restricted or repetitive patterns of behavior, interests, or activities. Deficits in social communication and interaction could include difficulty in making and maintaining relationships, understanding non-verbal communication, and failing to initiate social interactions. Examples of behavioral patterns that could indicate autism include an insistence on sameness and hyper- or hyporeactivity to sensory input.[1] Behavioral symptoms of autism have been found in children as early as 12-18 months.[2] Estimates vary, however, about one in every one hundred children has been diagnosed with autism.[3]

Measuring the severity of autism is based on these factors. Additionally, diagnoses note whether the individual has an accompanying intellectual disorder; these individuals also have a higher chance of having depression, anxiety, and motor deficits.[1]

There is a large amount of research done into autism, and new studies are continually being published.[4] On PubMed, for example, there were 7823 on autism published in 2022. Yet, there is a large need for this new research; little is known about the specific causes of autism, or about the relationship between autism and culture.[5] Autism research has typically focused on an Anglo-centric population, and this bias in research has resulted in some underrepresented minority groups reporting higher distrust of autism research.[6]

Culture, which can be defined as differences across different populations, plays a large role in health care
overall. Cultural beliefs can affect how people feel and think about their health, when they seek help for healthcare, and what symptoms are concerning enough to be reported.[7] In certain Asian cultures, the stigma associated with mental health disorders is often great enough that people believe their economic prospects, as well as their prospects of marriage, are weakened by being diagnosed. This in turn makes people with this set of beliefs less likely to seek help for a mental disorder.[8]

While internal cultural beliefs play a large role in people's relationship with healthcare, external factors also tend to impact how people view their health. Mistrust of medical professionals by underrepresented minorities arises from previous persecution and present-day issues surrounding racism and discrimination.[8] In the United States, for example, research has shown that Black and Hispanic minority populations, which have been historically oppressed, report higher levels of provider distrust.[9]

In addition to cultural beliefs, genetics can also shape differences in healthcare across cultures. It is important to note that it is generally agreed upon by the scientific community that the term race is a social category, and that the socially structured categories of race do not have a genetic basis.[10] A research study compared genetic variation between established races to genetic variation among these races, and ultimately concluded that there was no biological race.[11] Research from the National Human Genome Research Institute supports this finding.[12] Race, however, still plays a large role in people's ability to seek healthcare, both for the cultural reasons mentioned above as well as the overall discrimination and systemic racism present in the healthcare systems of many countries, including the United States.[13] Similarly, in many European countries, migrants, especially those of different races, face inequality and discrimination when seeking healthcare.[14]

Conversely, a population's geographic ancestry can affect their genetic makeup in many ways, making people whose ancestors were from a single part of the world predisposed to certain genetic mutations or variations. A population's geographic ancestry is different from its "ethnicity," the definition of which has been shown to be ambiguous.[15] Geographic ancestry refers to the fact that human adaptation due to geographically dependent natural selection has led to instances of "parallel divergence events."[16] Essentially, environmental factors make certain traits more or less desirable in different geographic contexts, resulting in natural selection for certain genes that then become favored by the surviving population in a given geographic region.

This review seeks to examine the differences in the autism diagnosis rate of different racial and geographical populations worldwide. It will discuss the influence of systemic biases, internal cultural influences, and parent and caregiver influences on the diagnostic process and tools used to diagnose autism. It will also examine genetic and biological differences that can affect disparities in autism diagnoses. This review will conclude by emphasizing the importance of timely and equal autism diagnoses and by discussing the need for further research and structural change.

Diagnostic Processes and Tools

The American Academy of Pediatrics (AAP) recommends that children should be screened for all developmental disorders during regular doctor visits at ages 9, 18, and 30 months, with autism-specific screening administered at 18 and 24 months. Parents, who have been reported to be good sources of information about their children, fill out a questionnaire, which is then screened by clinical staff. Parents have been reported to be good sources of information about their children.[17]

The United States Preventative Task Force, an independent panel of experts that makes recommendations for clinical practices, on the other hand, concludes that there is not enough evidence to either recommend or not recommend autism screening.[18] Interestingly, there are no international guidelines on autism diagnoses, and many research papers cite the AAP guidelines, regardless of what countries the research is conducted in.

There are issues with this diagnostic system, and different groups are disproportionately benefiting from the current diagnostic tools. In the United States, for example, studies consistently show that Latinx and Black children are diagnosed with autism later than White children; black children are, for example, diagnosed on average three years after their parents first express concerns.[19]

Providers and Healthcare Workers

Currently, autism diagnoses rely heavily on healthcare workers making a diagnosis based on their subjective experience with a patient. Unfortunately, this means that for all patients, a lack of confidence among doctors can serve as a barrier to correct implementation of the recommended screening procedures.[20] Additionally, an overall shortage of trained professionals means that there are less people who can actually conduct the screenings and diagnose autism,[21] with communities of color relying on Medicaid being impacted most drastically.[22]

Communication with families about symptoms may be impacted by provider implicit bias, an unintentional and automatic form of bias. This implicit bias could then impact the healthcare professionals screening or assessment of symptoms, which could in turn affect the diagnosis they ultimately give. For example, the stereotype and resulting bias that children of color more often have behavioral problems can lead to healthcare providers writing off autism symptoms as behavioral difficulties.[23] In fact, studies have shown that in the United States, African Americans are 2.6 times less likely to receive an autism diagnosis than white children.[24] Additionally, research has suggested that when some diagnosers observe “cognitive impairment” in “traditionally underserved minorities,” they are potentially less likely to recommend further assessment.[25] However, the correlation between being less likely to recommend this further assessment and provider implicit bias has not been further researched.

Language Barriers

In healthcare overall, language barriers have been shown to harm the patient-provider relationship and lead to lower levels of patient satisfaction, as well as decreasing the quality of patient care.[26] This problem is especially relevant in autism, as the screening process requires constant, consistent, and clear patient-provider communication in order to correctly understand a patient’s symptoms and arrive at a correct diagnosis. Indeed, research suggests that language barriers could harm parents' abilities to access health care services for children with autism.[27] In a study conducted in the United States, Hispanic and Latinx caregivers noted that information was not always provided in Spanish, while Korean-speaking participants specifically stated that language barriers and a lack of Korean-speaking professionals diminished the quality of care they received. The study also found that certain participants believed that their child’s diagnosis was delayed by healthcare providers who attributed their patient’s language delays with limited exposure to English.[28]

A cross-cultural study of autistic traits in India, Japan, and the United Kingdom examined the use and effectiveness of the Autism Spectrum Quotient (AQ), an autism diagnostic originally developed in the United Kingdom. The study aimed to discover common ‘key indicators’ of autism across different cultures; one of its many conclusions concerned language barriers. In order for the AQ to be administered across different cultures, it was also translated from English to Japanese, Hindi, and Bengali. The results of the study showed different levels of effectiveness across the different countries, and one of the potential causes this study offered was the translation. Specifically, the fact that words have varying connotations and are associated with varying ideas in the different languages; for example, the Hindi and Bengali translations of “spontaneous,” which appear in the survey, are more commonly used in written language than in spoken language. Therefore, parents’ responses to these questions may reflect their unfamiliarity with the word.[29] While this is not the only possible reason for the varying success rates of the AQ, it could certainly be an important factor.

Internal Cultural Beliefs

Another one of these factors, also addressed in the study, is different cultural expectations. As mentioned earlier, the cultural beliefs associated with different races and/or geographic populations can be an important factor in people's relationship to healthcare. Specifically, these beliefs can impact what people believe is worth consulting with and/or reporting to a doctor about. The same cross-cultural study (which looked at the efficiency of the AQ) suggested another possible reason for the varying success rates of the AQ: cultural norms. The study suggests that certain traits—spontaneity, for example—are encouraged or rejected to different degrees in different countries. The question “s/he enjoys doing things spontaneously” was a good indicator of autism in the United Kingdom, but not in India or Japan. In all three countries, 91%-97% of the control group reported enjoying spontaneity; in the United Kingdom, two thirds of the autism group reported disliking spontaneity, while less than one third of the autism groups in India and Japan reported the same. The study suggests that because Japan is characterized as a high uncertainty avoidant society, spontaneous activity is more structured, so few children object to the activity. For Indian children, the study offers the explanation that a prevalence in an authoritarian parenting style results in a general reduction of spontaneity, which makes it a less accurate indicator.[29]

A study done in Israel compared the differences in autism diagnoses among the Bedouin and Jewish populations in Israel, and reached similar conclusions. The study found that there was a large disparity in the percentage of children that finished the diagnostic process: 57.1% of Bedouin children completed the process, compared to 84.4% of the Jewish children. The study also found that Bedouin toddlers are approximately three times less likely to start the diagnosis process than Jewish toddlers. The study suggested a potential reason for this was that some of the questions on the test they were using, the Modified Checklist for Autism in Toddlers (the M-CHAT), were not relevant to the Bedouin population. An example of this type of question would be “does your child enjoy playing peek-a-boo/hide-and-seek?” [30]

This section discussed the influence of providers and healthcare workers, language barriers, and internal cultural beliefs on diagnostic processes. This review will now discuss varying genetic and biological factors that could affect autism diagnosis.

Genetic and Biological Differences

Research has currently identified over 100 risk genes for autism. According to a study done by Mount Sinai, these genes can be categorized in three main groups, including gene expression regulation (58), neuronal communication (24), and cytoskeletal organization (9).[31] The SFARI Gene Database, an extensive catalog of the genes associated with autism which ranks how related a gene is to a risk of autism, has identified 232 genes that are “clearly impacted” in autism. It has additionally identified 287 mutations that are associated with an elevated risk of autism as well as “linked to additional characteristics not required for an autism diagnosis.” It has further identified 702 genes that are strong candidates for being impacted in autism, and 112 genes that have “suggestive evidence.”[32]

Heritability is a measure of “genetic influence on variation.” If a trait or disorder has high heritability, its variance across a group of people or population is a result of, or can be explained by, genetics. Conversely, the variance of traits with low heritability cannot be explained by genetics and instead is often viewed as a result of environmental factors.[33] Research has not reached a conclusion on the heritability of autism. While many studies agree that heritability does play a sizable role in the development of autism, the actual heritability estimate varies considerably; a meta-analysis published in 2015 found that estimates of the heritability of autism were anywhere from 64% to 91%.[34]

A study performed in the 2000s based on research conducted with a Swedish cohort of siblings and published in 2017 suggested that the heritability of autism is about 83%. It included the note that this estimate could potentially be lower than what the cohort actually suggests as a result of the methods used.[35]

It is additionally worth noting that most genomic research occurs in populations of European ancestry, who do not reflect the global population.[36]

Variance in Single Nucleotide Polymorphisms

Pathway analysis (PA) is a set of commonly used tools that are supposed to help analyze biomedical data. It has many applications in both biomedical and physiological research, and it has helped in the identification roles of candidate genes. PA has been used in autism research in an attempt to figure out what genes play a notable role in the development of autism. Using previously identified genes, a study conducted in 2014 tried to find groups of gene mutations called SNPs that impact cellular pathways associated with autism and to apply machine learning to identify SNPs to make a “predictive classifier” for autism diagnosis. The results of this study were notable specifically because they referenced the different effect of the SNPs on different geographic populations; the subgroups the study looked at were Utah residents with Western and Northern European ancestors (CEU), Tuscan Italians (TUI) and Han Chinese.

The study developed a classifier based on the SNPs that they identified and was able to predict autism with high frequency in the CEU and TUI cohorts (85.6% and 84.3%, respectively). These accuracy rates are quite high. The classifier, however, was not as successful in predicting autism in the “genetically distinct” Han Chinese population; the paper itself described the classifier’s success as “sub-optimal.” Interestingly, the study cites the differing number of SNPs shared across the different populations to be a potential reason for this variance in accuracy. Indeed, according to the study, 627 SNPs differed between the TSI and CEU cohorts; this figure increased to 116,753 SNPs between the CEU and Han Chinese cohorts. The study goes on to suggest that more research can be conducted into SNPs for the Han Chinese population specifically, which could enable a more precise study of how SNPs affect different geographic populations.[37]

Vitamin D Exposure

A review published in 2011 took a different approach to studying the effect of geographic population on autism. It used a hypothesis that theorized that insufficient levels of vitamin D is related to developing autism. The study aimed to compare rates of autism in studies that differentiated maternal immigrant status and where the patient was born. It found that rates of autism increased in children of immigrant mothers compared with those of native-born mothers. Specifically, location of birth outside of Europe and America was a significant risk factor in studies conducted in Northern European countries; maternal birth outside of Australia was a significant risk factor for the study conducted in Australia. This is in line with what the study hypothesized they would find, as the researchers believed that based on genes affected by geography, people need different levels of vitamin D, and that by moving, the amount of vitamin D they need is no longer proportional to the amount of vitamin D they receive.[38]

This particular study does have several limitations that are worth considering. Notably, the study used skin pigmentation as a proxy for vitamin D levels. Since there is not a perfect correlation between skin color and vitamin D levels, using skin color as a proxy for vitamin D introduces a significantly higher level of error.[39] Additionally, there is no longer a direct correlation that can be found in the research between vitamin D levels themselves and autism diagnosis. As of a review published in 2021, there is still no conclusive data that proves that Vitamin D is directly related to autism, however, children with autism consistently report lower levels of vitamin D than neurotypically developing children.[40] Despite this, there is an argument that this study still has important results, and that its framework can be used in future research.

Urban vs Rural Environmental Factors

Studies have found an increased risk of autism in children who have been born in or grown up in urban settings as compared to a more rural environment. A study published in 2015 is an example of such research; it found that at birth as well as in childhood, a higher degree of urbanization resulted in a higher risk of autism. This same study also found a trend “towards a higher risk of autism for children changing level of urbanicity [the degree to which a particular area is considered urban] between birth and the end of follow-up compared to children with the same urbanicity level of residence at birth as current residence.” An exception was children born in the highest level of urbanicity (typically in the capital city) who had a marked decreased risk for autism if they moved to a lower level of urbanicity after birth.[41]

The reasons for this variance are debated and likely very complex. Urbanicity may be a proxy for other risk factors, such as air pollutants and noise levels, though there is little actual evidence to support this.[41]

Another possible reason for this difference is that urban areas, which tend to be more densely populated, have greater autism awareness. A study published in 2017 suggests that the “geographic circumstances” of living in a rural area, as well as the decreased availability of diagnostic services, result in the the primary care system's reliance on other institutions (ie. schools) for autism-related care. The same study goes on to suggest that the comparative lack of autism awareness in rural communities, which exists on community, school, and healthcare wide levels, result in less diagnosis.[42] If this were true, it would be an example of how the intersection between the effect of geographic location as well as the autism diagnostic process can affect overall autism diagnosis.

Discussion

Importance of Early Diagnosis

Equitable and timely autism diagnoses are vital in ensuring the best possible care and prognosis for all autistic individuals. For children with autism, intervention is heavily focused on the development of social skills and awareness, as well as on "habilitative therapies," which are therapies focused on improving quality of day-to-day life. Together, these therapies can help optimize the developmental path of an autistic individual. Difficult-to-correct behaviors or habits are easier to correct the younger the child is, making it important to diagnose the children as early as possible to give them the most time to respond to and adapt based on the therapies that are used for them.[43]

For example, a study published in 2016 supported the clinical value of Parent-mediated social communication therapy for young children with autism, or PACT. PACT was a randomized controlled trial that studied parent-mediated social communication intervention for children. The study followed up on the results of the therapy and found that it did work. This therapy, however, was initially conducted on children aged two to four years old; this age window shows the importance of early diagnosis—children would only be eligible for this therapy if they were diagnosed at latest by two years old, a relatively young age. Having strong diagnostic tools and equitable access to diagnoses would therefore help more children be eligible for care.[44]

Additionally, diagnosing autism early can help families better plan for taking care of a child with autism. This includes trying different therapies, as well as looking into different schools or programs that can better support children with autism.[45]

Remedying the Language Barrier

One of the most important ways to ensure more equitable diagnoses for all people with autism is by fixing the language barrier issues that have been studied and proven to exist in autism testing. Specifically, as mentioned above, questionnaires are often written in one language, often English, and then translated after the writing process is complete. While this process does effectively create a diagnostic in its original language, the meaning of the words can be lost or changed when they are translated. Further, specific words have different connotations in different cultures, as shown with the word spontaneous and the question about playing hide-and-seek described above.

To solve this, a potential solution would be to write the original questionnaires in several languages. While the writers would work together to ensure that the tests are overall assessing the same qualities, they would write the diagnostics in different languages, ensuring that the words chosen accurately convey the intended meaning. By not making only one language the original and then translating from that model, and instead creating several "original" translations with potentially different words but more similar meanings, researchers and writers can ensure that the tests being created are more fair and inclusive. This increased inclusivity could affect autism diagnoses by making the diagnostic more accurate, which could in turn increase the speed of the diagnosis and lower the chances of one or several incorrect diagnoses, which have detrimental impacts on the patients and their families.

Additionally, involving writers who speak different languages increases the chances of more diversity in the group of people who write any given diagnostic. This is helpful because a large part of the language barrier issues stem from cultural customs referenced in the questions (like hide-and-seek). These traditions are important to understand, especially because autism diagnoses are heavily reliant on whether the patient and their caregivers understand the questions being asked in the context of their culture. Having more people involved from the start in the writing process means more perspectives and more cultures included in this process, which allows the diagnostics to ultimately have increased awareness for these cultural differences, which would then result in the diagnostics' relatability to a particular group of people.

Healthcare Provider Awareness and Diversity

In order to then effectively administer these diagnostics, health care professionals and providers also need to have more training on how to interact with children with autism, as well as with their families. As mentioned above, there is a shortage of healthcare providers trained in diagnosing autism; this disparity is especially clear considering that, in general, Black, Hispanic, and Native American people are underrepresented across several health care professions.[46] According to the American Association of Colleges of Nursing, for example, 80% of the registered RN personnel are white.[47]

These disparities need to be addressed for several reasons. First, healthcare workers can often misconstrue actual language developmental delays with not understanding the English language. Having a more diverse healthcare staff to administer the diagnostics could help solve this issue, as a more diverse staff who speaks different languages could help distinguish between the two possibilities. Additionally, the patient-provider relationship is very important in all of healthcare, and is incredibly vital in the diagnosis and treatment of autistic children, as the providers need to know patients well in order to understand and diagnose them. Research suggests that people feel more comfortable with a healthcare provider of their same race, which overall emphasizes the importance of having diversity—it would ensure that the patients are as comfortable as possible. This research then also suggests patients are more likely to adhere to a physician's recommendations if they are of the same race, which is important for families deciding what therapies to use or further diagnoses to seek.[48]

It is also important that all healthcare workers, regardless of race, have a high level of training in autism diagnosis and care. Because diagnosing autism requires a trained professional, it is vital that more providers are trained in making this diagnosis. Currently, only highly trained doctors or child psychologists can diagnose autism, however, by making the training more accessible to other healthcare professionals, like nurses, a larger overall group of qualified providers would be able to diagnose autism.[14] This would also make autism diagnoses more accessible, because there would be more personnel capable of making the actual diagnosis, in communities where it is currently harder to access, for example in more rural areas.

Additionally, standardizing the guidelines for autism screening and intervention would largely benefit healthcare workers, as well as the families they care for. Currently, there are no international standards for autism testing, as well as what diagnostic should be used, or what the most effective autism interventions are. The first step to solving this would be to have international guidelines for autism screening, ideally released by the World Health Organization or an equivalent institution.

It would also be necessary for individual countries to release their own guidelines that take into account their specific circumstances and resources, as well as cultural differences that could affect the autism diagnoses, such as the screening tool that makes the most sense for that culture. In order for these guidelines to be evidence-based, further research is needed.

Further Research

As with all topics in science, further research is needed; this is especially true with autism, as little is currently known about its causes or long term effects. While more research needs to be done into these causes of autism generally, as well as the overall long term effects, some new research should specifically focus on studying non-White people, as well as people not of European descent. As seen in the study that used SNPs in different subgroups, researchers have already figured out the framework for models that can be used for other groups—in that particular study, researchers focused their model on the SNPs of the CEU cohort, but recalibrating their diagnostic to focus on the Han Chinese SNPs, for example, could provide a tool that is more accurate that the one produced by the current research.

Additionally, several methods from the study on Vitamin D study can be used—although this particular study used skin pigmentation as a proxy for Vitamin D levels, studying the actual Vitamin D levels in patients could lead to conclusive evidence about whether vitamin D levels themselves have anything to do with autism. This could then lead to discoveries about the role of immigrant status in children developing autism.

Resources and Awareness

Lastly, it is imperative that awareness about autism continues to be increased around the world. Despite decreasing in the past years, stigma around autism and other neurodivergent disorders still exists. It is vital that this stigma is combated and awareness is promoted, as a lack of awareness harms the person with autism, as well as their family. For a child, increased awareness can help them fit in in varying social settings without feeling left-out or excluded, which can ultimately benefit their mental health. For an adult, being more aware about the symptoms of autism can help them figure out if they should seek a diagnosis—while for adults, it may be too late for some intervention therapies, having the correct information is always beneficial. For the families and caregivers of someone who has autism, especially a child, being well-informed is incredibly important, and it allows them to make well-informed and helpful choices that will benefit the patient.

Another reason that autism awareness is important is because increased awareness leads to more resources being given to communities where access to diagnosis and care for people with autism is lacking or less than sufficient. For example, increasing autism awareness in rural communities could help more people gain accurate diagnoses. Further, increasing awareness of the fact that people in rural communities do not have access to the appropriate tools could urge larger foundations and organizations to donate or send more resources to these places.

Conclusion

This review has examined differences in the rate of autism diagnosis in different racial and geographical populations worldwide. It has also discussed the influence of systemic biases, internal cultural influences, and parent and caregiver influences on the diagnostic process and tools used to diagnose autism. It further looked at genetic and biological differences that can affect disparities in autism diagnoses. This review then emphasized the need for a timely diagnosis for autism and offered solutions for the language barrier and a lack of provider diversity. This review will conclude by discussing the need for further research and further awareness.

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