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Enhancing Early Detection: Improving Autism Spectrum Disorder Diagnostic Processes

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Abstract

Early identification and intervention of Autism Spectrum Disorder (ASD) has been well-established as extremely important for developmental outcomes, as the most efficacious treatments for the disorder occur prior to five years of age (McCarty & Frye, 2020). In this paper, divided into two chapters, I examine potential changes to the ASD diagnostic process to aid the goal of early intervention.

In Chapter 1, I discuss how modifications to current gold-standard diagnostic tools could help with this goal. Efforts to improve early detection and intervention for Autism Spectrum Disorder (ASD) have led to the development of screening and diagnostic tools such as M-CHAT-R, ADOS, and ADEC. However, despite the progress in this field, the average diagnostic age remains significantly older than optimal (McCarty & Frye, 2020). This literature review delves into the limitations of existing tools and proposes modifications aimed at enhancing early detection, such as revising scoring systems, incorporating assessments for motor issues, and improving cohesion between screening and diagnosis.

In Chapter 2, I expand upon how utilizing the motor domain in the ASD diagnostic process could aid early detection. While research has consistently demonstrated a link between ASD and delays in the motor domain, the mainstream screening practices often neglect motor considerations. Specific trends in infant motor development, such as balance (Odeh et al., 2020), gait, and postural control (Fulceri et al., 2019), have been identified as predictive of ASD diagnosis. Moreover, motor difficulties have been found to impact outcomes in domains core to ASD itself (Libertus & Violi, 2016; Iverson et al., 2018). Yet, less than 1% of individuals with ASD receive clinical recognition for motor impairments, and even fewer receive targeted therapeutic

interventions (Bhat, 2020; Licari et al., 2019). Incorporating motor assessment into current screening practices and modifying diagnostic labeling to include motor concerns could improve early recognition of ASD.

Chapter 1: Addressing the Shortcomings of Gold-Standard Autism Spectrum Disorder Screening and Diagnostic Tools

Addressing the Shortcomings of Gold-Standard Autism Spectrum Disorder Screening and Diagnostic Tools

Autism Spectrum Disorder (ASD) is one of the most prevalent neurodevelopmental conditions, with around 1 in 36 children meeting the criteria for diagnosis (Centers for Disease Control and Prevention, 2020). ASD is often marked by persistent difficulties with social communication, interaction, and repetitive patterns of behavior, but presentations of the disorder vary greatly across the spectrum. In many cases, ASD can result in clinically significant impairment in social, occupational, and other important forms of functioning (American Psychiatric Association, 2022). Research has suggested that early diagnosis and subsequent intervention can lead to favorable outcomes (Ribeiro et al., 2022; Wergeland et al., 2022), thus much research has been put into developing risk screening and diagnostic tools based on early behavioral correlates of ASD. In this paper, I will examine three tools in particular: the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R), the Autism Diagnostic Observation Schedule (ADOS), and the Autism Detection in Early Childhood (ADEC). I will overview how these tools are administered and how they fit in the field of ASD diagnosis, followed by an investigation of some possible shortcomings, including issues with appropriate sensitivity, difficulties with the heterogeneity of the disorder, and asymmetry of the low-end range that these tools can be administered (Kuhfeld & Sturm, 2018; Ribeiro et al., 2022), all of which hinder the goal of diagnosing the disorder and providing intervention as early as possible. With these shortcomings in mind and with the additional conclusions of more modern ASD correlational research (Neimy et al., 2017; Tye et al., 2020; Wagner et al., 2020; Winder et al., 2013; Miller et al., 2021; Roberta et al., 2021; Iverson et al., 2019), I will propose some potential modifications to

these tools, combining them into a single, comprehensive tool for ASD screening and diagnosis. I will then discuss potential future directions to aid the goal of lowering the diagnostic age, in addition to the continued need for evidence-based interventions that would follow diagnosis.

Overview of Autism Spectrum Disorder

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition that can cause developmental differences in numerous domains, most notably manifesting in social, communicative, behavioral, and motor challenges. In many circumstances, ASD can have a debilitating impact on individuals and their families. Communication difficulties, social barriers, and impulse control issues often associated with ASD can make even simple daily activities and skill acquisition significantly more difficult than they would otherwise be for someone with neurotypical development. As children with ASD develop into adulthood, they may have difficulty creating and maintaining friendships, communicating with those around them, and struggle with independence, often making it more difficult to pursue meaningful education and employment (American Psychiatric Association, 2022).

ASD can also result in additional difficulties as it is highly comorbid with other disorders. Research has consistently shown that individuals with ASD are at a far greater risk of developing depression and ADHD (Centers for Disease Control and Prevention, 2023). Additionally, around 40% of those with ASD also struggle with anxiety, which can be particularly challenging as it can exacerbate many common symptoms of autism, such as issues with social interaction, communication, and sensory sensitivity (Zaboski & Storch, 2018).

Importance and Difficulties of Early Diagnosis

ASD can have such debilitating effects at such an early age, often leading to hindered development during important formative years. As such, early detection and subsequent diagnosis are very important for allowing the introduction of timely and effective therapeutic intervention strategies. In young children with ASD, their neuroplasticity is a very positive asset when it comes to the improvement of outcomes (Ribeiro et al., 2022). The earlier a diagnosis is made, the sooner treatment can begin to help work on skills and abilities that would otherwise go undeveloped without intervention. The potential for positive synaptic changes becomes increasingly limited as the brain ages out of infancy, making the science of early detection and treatment of ASD one of paramount importance (Ribiero et al., 2022). In fact, analysis of intervention strategies in both research and clinical settings has shown that behavioral interventions only show efficacy in children aged five years or younger (Wergeland et al., 2022). Some studies have placed the age important for intervention even earlier, as there has been an observed noticeable decline in interventional efficacy even after a child turns three years old (MacDonald et al., 2014). This can even complicate research design for clinical studies concerning ASD interventions, as there can be potential ethical concerns with assigning a young child with ASD to a control group during these important developmental years, thus limiting their treatment during peak plasticity (Wergeland et al., 2022).

Barriers to Simplicity of Diagnosis

Although ASD is one of the most prevalent developmental disorders in the United States, with about 1 in 36 children meeting the criteria for diagnosis (Centers for Disease Control and Prevention, 2020), ASD is also defined by its heterogeneity, that being its diversity of presentation, often making it difficult to diagnose. Because it impacts such a

wide array of domains, symptoms and challenges may differ greatly from one affected individual to another. This immense variability makes it quite difficult, arguably impossible, to create all-encompassing diagnostic criteria. What may be indicative of ASD in one individual may not be in another, differences which may be further exacerbated by ASD's frequent comorbidity with other mood and behavioral disorders. Additionally, in many cases, the limitations of young children with ASD may not be readily apparent, especially in early developmental stages, as their immediate environment may not yet demand or expose the full extent of their challenges (Wergeland et al., 2022). To address these complexities, diagnostic tools need to be flexible, inclusive, and discerning, which is often a very delicate balance to strike. Unfortunately, the current mean age for obtaining an ASD diagnosis is 4 years and 3 months old, which falls well into the nebulous window where the efficacy of interventions begins to decline (McCarty & Frye, 2020). Even if we take the most generous assessment of the first five years of treatment being effective before a decline, many children on the older side of this distribution fall outside of that range. Thus, at this age, many children who acquire a late diagnosis may have an even greater difficulty developing the necessary skills to keep up with their neurotypical peers. As such, reducing the average age of a diagnosis is of utmost importance for the subsequent outcomes of affected children. Additionally, low socio-economic status is a significant predictor of late ASD diagnosis (Emerson et al., 2016), making this goal important for increasing the equitability of healthcare in general.

Current Early Screening and Diagnostic Strategies

Although the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition, Text Revision (DSM-5-TR) is the ultimate reference for the diagnosis of ASD, it

only lists the symptomatic criteria for diagnosis; no standardized, formal test for ASD exists in the DSM-5. Thus, numerous types of instruments have been developed to attempt to fulfill this role. These tools have taken many forms, including checklists, questionnaires, interviews, and direct observational sessions. The diversity of these tools means that they often serve different purposes on the path to diagnosis. Some tools are very brief and are meant to simply find children who may be at risk for developing ASD. In contrast, others are more rigorous and are meant to screen children already referred for behavioral or developmental concerns to the point of clinical diagnosis. This distinction has led to screening tools being split into two separate categories: Level 1 and Level 2. Level 1 screening tools are often used in broader healthcare settings, and as mentioned above, are often quite brief, making them very accessible and useful for parents' and clinicians' initial risk assessment. In contrast, Level 2 screening tools are more comprehensive and rigorous and dive deeper into more specific concerns and symptoms of ASD. Level 2 tools are valuable in that they provide a far deeper, and often individualized view of a child's particular situation (Nah et al., 2014). Of these tools, I specifically examine the following: M-CHAT-R, ADOS, and ADEC, as they are some of the most frequently used. Taking a look at the most heavily utilized tools is important, as their reach and impact is far greater.

Level 1: M-CHAT-R

The Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R) is one of the most frequently utilized Level 1 tools for ASD screening, both in the United States and internationally (Robins et al., 2014). It is designed and has been validated to be used for children aged 16-30 months old. It consists of a series of 20 yes or no questions to be filled out by a parent or caregiver in regard to a child's social, communicative, and

motor abilities (for example, “When you smile at your child, does he or she smile back at you?” (Robins, 2023)). It can be completed in just a few minutes, making it very accessible, as Level 1 screening tools tend to be. The scoring manual suggests that a score of 0-2 represents a very low risk, a score of 3-7 represents a moderate risk, and a score of 8-20 represents a high risk of ASD (Pop-Jordanova & Zorcec, 2021). There is also a follow-up module, the M-CHAT-R/F, with additional questions meant to be filled out after a moderate risk result. One goal of the M-CHAT-R is to maintain a very high level of sensitivity, meaning that it attempts to detect as many potential cases of ASD as possible: in a sample of nearly 19,000 toddlers, 98% of those flagged as at-risk presented with developmental delays or concerns, and 54% went on to be diagnosed with ASD (Robins et al., 2014). An analysis of the M-CHAT-R found that around 93% of children fell into the low-risk category, 6% of children fell into the moderate-risk category, and 1% of the children fell into the high-risk category (Robins et al., 2014). These numbers result in a rate of detection of 1 in 127, which does not line up with the CDC’s 1 in 36 figure (Centers for Disease Control and Prevention, 2023). This could perhaps be explained by the fact that some cases of ASD are only detectable after more peer interaction can be observed in schools, which can often come after the M-CHAT-R’s administration, but it is an interesting asymmetry nonetheless. Overall, the M-CHAT-R is thought to be one of the most effective Level 1 screening tools in many ways; the simplified scoring paired with specific algorithms based on outcome makes it a very comprehensive assessment.

Level 2: ADOS

The Autism Diagnostic Observation Schedule (ADOS) is considered a ‘gold standard’ Level 2 assessment for the clinical evaluation and diagnosis of ASD. The

ADOS involves an in-person observational session where a trained clinician observes and assesses a child's communication, reciprocal social interaction, creativity, stereotyped behaviors, and restricted interests by giving them a set of semi-structured tasks. The ADOS consists of five modules, each of which is designed to be administered to individuals based on their functional language level, ranging from toddlers (Toddler Module) to adults (Module 4) (Kuhfeld et al., 2018). The Toddler Module, specifically, is the most recent update to the ADOS, made to address issues that it was ineffective and unreliable for children younger than 30 months of age. This module utilizes the same style as other ADOS modules, and targets language and communication, reciprocal social interaction, play, and stereotyped/restricted behaviors. With the addition of the Toddler Module, the low-end effective age limit of the ADOS is officially 12 months (Luyster et al., 2009). As a gold-standard tool for ASD diagnosis, the ADOS has proven to have effective interrater reliability: a review of the tool found that agreement in diagnostic classification between different administrators of the test ranged from 92% to 98% in Modules 1 through 3 and from 87% to 97% in the Toddler Module. Additionally, the ADOS's predictive validity is also strong for a disorder as heterogeneous as ASD, with the sensitivity of Modules 1-3 ranging from 60% - 95% and specificity ranging from 75% - 100%. Both sensitivity and specificity for the Toddler Module were at or above 86% (Lord et al., 2012). To this day, the ADOS has become a critical component of both research and clinical practice surrounding the diagnosis and subsequent intervention of children suspected of having ASD, and it is evident that it changes and updates along with the direction and needs of the scientific community.

Level 2: ADEC

Although not considered a 'gold-standard' tool like the ADOS, the Autism Detection in Early Childhood (ADEC) was created to address some of the shortcomings of other Level 2 screening tools, particularly the low-end age limit of their effectiveness. The ADEC is specifically designed to detect ASD in children between 12 and 36 months of age. It is composed of 16 discrete behaviors that are thought to reflect core high-risk behaviors that can be identified in children in the this age range. Each item of behavior is operationalized to limit ambiguity between administrators of the screening tool. Unlike many other tools, the ADEC has a scoring system of partial credit where each behavior is given multiple trials to see if the child is able to succeed in none, some, or all of the trials. For example, one measured behavior, social response, is operationalized as whether or not a child responds to their name at all over the course of five separate trials. If the child responds in the first two attempts, they are scored a 0 on this unit (a lower score implies a lower risk of ASD); if they respond within the next three attempts, they are given a score of 1; if they do not respond at all, they are given a score of 2. This is an attempt to address the heterogeneity and sometimes spontaneity of certain traits associated with ASD, something that is frequently ignored in other tools (Nah et al., 2014). Again, although the ADEC is not considered gold-standard like the M-CHAT-R or the ADOS, it includes useful features that strike a middle ground between the accessibility of the former and the rigor of the latter.

Limitations of Current Screening and Diagnostic Tools

Overview

Early diagnostic tools for ASD represent an ongoing area of research and development, as the science around the disorder continues to grow. While significant

strides have been made within the recent decades, there are still some weaknesses and blind spots in all diagnostic tools, and the three aforementioned ones are no exception.

Sensitivity and Specificity

Under the context of ASD screening tools, the value of sensitivity versus specificity is a critical consideration. Sensitivity refers to the ability of a tool to identify individuals with ASD, limiting cases that slip through the cracks, or false negatives. Conversely, specificity refers to the ability of a tool to prevent cases of people without ASD meeting the criteria of their measure, or the limitation of false positives. Balancing between these two can be very difficult, as it is easy to inadvertently decrease one while attempting to increase the other, and vice versa.

For example, one study found that the M-CHAT (the iteration of the tool that preceded the M-CHAT-R) struggled with specificity in a sample of 18-month-old children, and had issues with a poor positive predictive value (PPV) of 0.36. This PPV implies that only 36% of those that screened positive were later actually diagnosed with the disorder. The study speculated that this may have been a result of the M-CHAT's structure of only yes or no questions, leaving very little room for nuance (Sturner et al., 2017). Although the revised M-CHAT-R removed three items that were culprits of generating false positives, the binary structure still remains. One might argue that specificity should not be an issue with a Level 1 screening tool, as raising awareness of potential developmental red flags is the goal, not official diagnosis. However, a false positive can cascade into other negative effects, such as unwarranted parental anxiety and longer appointment wait times for secondary screening for children who actually have ASD. Conversely, a trade-off of increasing the specificity and decreasing the sensitivity might ultimately lead to fewer children being identified who go on to develop

ASD as well as missing out on children with other developmental disabilities with equally valid needs of early identification (Schjølberg et al., 2021).

Research has also shown that the ADEC can also struggle with specificity. In another study, sensitivity of the ADEC ranged from .93 to .94 and specificity ranged from .62 to .64 when applying a cutoff score of 11, which corresponds to the ADOS's recommendation of moderate risk for ASD. An improved balance of sensitivity (.85 - .87) to specificity (.79 - .82) was achieved using a higher cutoff score of 14, which corresponds to the ADOS's classification of high-risk of ASD. Even with the higher sensitivity, some of the false negatives revealed other flaws of the ADEC. One child who scored at low-risk for ASD, but was later diagnosed was characterized as having a typical, if not above-average developmental level but poor social skills. It was suggested that social limitations are not evident enough before preschool or other social situations are encountered (Hedley et al., 2015).

Heterogeneity and Difficulty of Diagnosing Youth

Although it is considered a gold-standard measure, the ADOS is also not without fault. Some research has shown that the ADOS struggles to account for the heterogeneity of ASD, which is certainly an issue since this is such a hallmark feature of the disorder. In one examination of its precision, the ADOS was shown to be less effective at assessing mild cases of ASD, where clear-cut symptoms may not be as typically present. Additionally, the ADOS was found to be less effective at taking note of restrictive and repetitive behaviors, another marked sign of ASD (Kuhfeld & Sturm, 2018). This shortcoming is one that might be even more important for early diagnosis, as these behaviors can appear before a child enters social scenarios with peers, and thus could be spotted early if better accounted for in the ADOS.

Underrepresentation of Motor Limitations

Additionally, one notable limitation in almost all commonly used ASD screening tools is the lack of criteria that incorporate motor difficulties into the assessment process. While ASD is primarily characterized by challenges in social communication and repetitive behaviors, motor difficulties are frequently observed in individuals with the disorder. Despite studies that have estimated the proportion of those with ASD and co-occurring motor difficulty at over 80% (Bhat, 2020), the diagnostic criteria in the DSM-5 neglect to include anything regarding motor impairments (American Psychiatric Association, 2022). These difficulties can manifest as issues with coordination, motor planning, and fine and gross motor skills. Neglecting to include motor difficulties in screening criteria may result in the underrecognition of individuals with ASD who exhibit these challenges. Research has found that some motor impairments in those with ASD are present at birth, so the lack of inclusion of this domain in diagnostic criteria seems to be a potentially underutilized aspect of ASD (Neophytou, 2021). None of the M-CHAT-R, ADEC, or ADOS explicitly test for motor issues in their criteria, and instead, they strictly focus on the social, communicative, and behavioral domains of ASD. In Chapter 2, I will further examine the motor domain's absence from screening practices.

Clinicians' Experience with ASD Diagnostic Tools

Some research has also investigated the experiences of health professionals who use tools such as these to diagnose ASD. Two issues were raised that are relevant to the particular tools discussed, those being parental knowledge and professionals using their own judgment in cases of abnormal presentation.

The issue of parental knowledge being a strong variable factor is quite relevant for tools such as the M-CHAT-R, which is filled out in full by the parent or caregiver of the child in question. Although it may be difficult for the M-CHAT-R to account for this in its items, it is absolutely something to consider. A parent who struggles to correctly interpret the items may give inaccurate answers, and subsequently lead to a false positive or negative.

As for the issue of professional judgment, this is particularly relevant for tools that involve direct observation from a trained professional, such as the ADOS. Those who administer the ADOS often have extensive prior experience with individuals with ASD, and this is supplemented with clinical training provided by an independent trainer or the test publisher (Lord et al., 2012), however, this does not eliminate all interrater variance, especially in atypical cases. As mentioned earlier, the ADOS already struggles with abnormal presentations of ASD, so health professionals would attempt to overcome this by supplanting their own judgment in a situation. Although it is good that health professionals are aware of this weakness of the ADOS, this results in being left to make a judgment call, cascading into problems of false positives and false negatives based on one individual's opinion (Howes et al., 2021).

Gender Differences in ASD Diagnosis

Another shortcoming of current diagnostic tools is the fact that the diagnostic ratio of ASD is heavily skewed towards those assigned male at birth (AMAB). The factors underlying this difference are predominantly unknown, but beliefs that diagnostic tools are built and validated against samples of mostly AMAB individuals, thus making AFAB presentations of symptoms less represented in these tools is a concern (Beggiato et al.,

2017). Unfortunately, due to the sheer complexity of this issue, this problem with diagnostic tools is beyond the scope of this paper.

Contemporary Correlates in ASD Research

Although much research has been put into discovering correlates, the exact cause or set of causes of ASD remains inconclusive. Both genetic and environmental causes have been investigated, but with little to no success, creating a true causal link between a specific aspect of those domains and ASD. As a result, contemporary research has continued to investigate very early infant correlates of the disorder, not necessarily trying to find the root cause, but instead looking for features or behaviors that may indicate the potential of a future diagnosis of ASD. The heterogeneity of ASD can make this difficult, as very few, if any, behaviors are common to all presentations of the disorder, but research of this kind is particularly useful for building and enhancing these tools for screening and diagnosing. As this correlational research expands and makes new discoveries, it is important to consider updating diagnostic tools to incorporate the findings.

Interaction with Toys/Objects

One very common field of correlational research is to look at young children's patterns of interaction with inanimate objects, such as toys and other things that a child might be interested in. Research from this field has already been utilized in screening and diagnostic tools: the M-CHAT-R includes a question about whether or not the child frequently engages in 'make-believe' play with toys (Robins, 2023), and the ADOS heavily incorporates observation based on a child's interactions with various toys. However, recent research has found even more infant-object interaction patterns that

may predict later ASD diagnosis, some of which can be observed well before the M-CHAT-R's self-proclaimed low-end effective range of 16 months of age.

One study (Miller et al., 2021) found that unusual inspection of inanimate objects (prolonged visual inspection, squinting at objects, looking at objects from peripheral vision) is often present and stable in infants later diagnosed with ASD at as young as 9 months old, and can predict a reduction in social engagement just three months later. It is advised that close inspection of these behaviors could aid the goal of early ASD detection. Additionally, research has also investigated interactions with objects and an infant's ability to attentionally shift from a toy to relevant social information. In this study, 12-month-old infants were set to play with a toy together with an examiner, who would then appear to injure themselves. Infants that were later diagnosed with ASD were found to more frequently continue to focus on the toy than on the examiner when compared to neurotypical infants. Although this finding is not perfectly foolproof, of the 129 in the study, the 11 infants who demonstrated the highest rates of attention shifting were all found to have no developmental delays (Hutman et al., 2011), so this may be a useful tool for eliminating potential high-risk infants if they score well on this test.

The presence of studies such as these that find behavioral correlations in infants so early in development, even up to seven months prior to the low effective range of the M-CHAT-R, shows that there is potential for these tools to be updated so that they can be effectively administered even earlier.

Social Interaction and Facial Recognition

Another commonly researched area of investigation is that concerning infants' social interaction and facial recognition. Infants later diagnosed with ASD often exhibit distinctive patterns in social interactions and attentiveness to facial cues and

expressions. Similarly to interactions with objects, these behaviors also can appear very early in development. Since differences in abilities in communication and social interaction are among the most well-known aspects of ASD, current screening and diagnostic tools, including the M-CHAT-R, ADEC, and ADOS, heavily incorporate this aspect of the disorder in their questionnaires and examinations. However, as with all fields of ASD research, the research in this domain is nowhere near reaching a full consensus on when and how these behaviors develop.

However, research has shown that many of the infant behaviors often associated with later ASD diagnosis may be present even earlier than many of these screening and diagnostic tools claim. A study including data from over 14,000 parents assessed developmental concerns that they might have for their child or children (Neimy et al., 2017). Among the parents of children who were later diagnosed with ASD, the authors found differences in the infants' frequency of spontaneous social interactions and eye contact, among other correlates, as early as 6 months of age. These two behaviors are heavily featured in the M-CHAT-R, but similarly to interactions with inanimate objects, they appear far earlier than the Level 1 screening tool seems to suggest. Incorporation of this information could certainly allow for a reconsideration of the believed effective age range of the M-CHAT-R to better mirror a diagnostic tool like the ADOS Toddler Module, which is statistically validated for infants as young as 12 months (Luyster et al., 2009).

Another study found evidence that 13-month-old infants at a high risk of ASD initiate spontaneous communication at significantly lower rates than same-age infants with a low risk of ASD. Within this study, 3 of the 15 high-risk infants were later diagnosed with ASD, and upon re-investigation of the data, all three of the infants were

at or near the low end of the study's measure of spontaneous communication (Winder et al., 2013). Although the sample size of this study was relatively small, it is further evidence that these social correlates may be evident significantly earlier than 16 months of age.

Motor-Related Correlates

As mentioned previously, the current diagnostic criteria for ASD largely focus on social communication challenges and the presence of repetitive and restricted behaviors, and deficits in gross- and fine-motor abilities are not explicitly mentioned. However, there is a growing body of research that suggests that differences in these domains are significantly related to ASD risk. One such study found that high-risk infants had significantly greater difficulty completing gross-motor tasks, and even more specific than that, actions that required utilizing multiple body parts to counteract the effects of gravity and maintain balance on the move (this included pulling on an experimenter's fingers to a sitting position from a lying position, and reaching for a toy while on their stomach) were particularly notable in the high-risk group (Iverson et al., 2019). This study suggests that there is strong evidence for keeping an eye on very specific motor-related abilities, and could be a first step in including motor differences in a diagnostic manner.

Neuroscientific Correlates

Another contemporary field of ASD research involves investigating neuroscientific correlates. Unfortunately, brain imaging techniques can be expensive, making them difficult to make readily accessible, thus impractical to include in routine diagnostic procedures. As such, this domain is not included in any of the mentioned

screening or diagnostic tools, but that does not mean that they cannot help inform the development of these tools.

For example, one study investigated the neural processing involved in facial recognition in 8-month-old infants with and without ASD, and found that it is generally atypical in infants later diagnosed with ASD. The study claims that this represents a strong candidate for a predictor of later behavioral correlates of ASD that would later arise (Tye et al., 2022).

Another study looked to map early brain network development and efficiency in infants both at a low-risk and high-risk for ASD, first at 5 months and again at 10 months of age. The study found that when compared to low-risk 5-month-olds, the neural networks of high-risk 5-month-olds show an overgrowth in local functional connection, which the researchers speculate may not support efficient communication between different, more distant regions of the brain. Interestingly enough, these differences no longer existed when the infants came back for the scan at 10 months, which raises an interesting implication that perhaps some of these neurological differences only exist in very early development, but then disappear after they have impacted development in some facet (Zhang et al., 2022).

In summary, it may not be currently feasible to incorporate these neuroscientific findings into current clinical practices of ASD screening, but continued development in this field has the potential to pick up on high-risk candidates even earlier than they can develop the behaviors that would subsequently be associated with ASD. With the goal of early diagnosis in mind, this is a particularly interesting field due to its ability to potentially pre-date behavioral correlates.

Modified ASD Screener Comprising Other Tools (MASCOT)

With the combined understanding of the M-CHAT-R, ADEC, and ADOS' strengths and weaknesses, enhanced by a body of additional correlational research, I believe that some modifications can be made to these tools to better serve the goal of early diagnosis of ASD in infants. I propose a combination of these tools into a singular, multi-level screening and diagnostic tool, the Modified ASD Screener Comprising Other Tools (MASCOT) that would be able to incorporate the strengths of each while covering for some of their shortcomings. This tool would remove the asymmetry of the low-end age restrictions between the M-CHAT-R and the ADOS, creating a standardized age of diagnosis at 12 months of age.

Cohesion Between Both Levels

One primary change would be instead of sticking to the current clinical practice of distinct and independent Level 1 screening and Level 2 diagnostic tools, this Combined-Level tool would simply be referred to as a single, two-part tool. Information from the initial screening, which would take the form of a modified M-CHAT-R (MASCOT-L1), would be incorporated into the actual diagnostic session, which would be a modified version of the ADOS (MASCOT-L2). This change would serve multiple purposes all stemming from one major benefit: the data collected from the initial screening would be used to influence the conducting of the diagnostic session. For example, if the MASCOT-L1 flagged a child as having a moderate risk for ASD, and indicated that the child had significant concerns with items on the questionnaire that related to social interaction, but little to no concerns with items that related to repetitive or restrictive behaviors, the diagnostic session could be abbreviated to place more focus on the social interaction aspect and spend less time on observing for a symptom that

may not be relevant. This change would shift the MASCOT away from specifically attempting to diagnose cases of ASD but instead focus on identifying the domains of developmental delay that are associated with the disorder. Although a child may not display restrictive behaviors, and thus not qualify for the diagnostic label of ASD, allowing them an assessment to address their potential developmental concerns is equally as important as a child that displays traits in both domains. It would allow for a more detailed investigation of the particular domains of concern, and could also provide the added benefit of reducing overall time to administer the MASCOT-L2, as diagnostic sessions could be shorter if redundant parts are abbreviated.

Additionally, the current M-CHAT-R has a low-end effectiveness of 16 months, but it is important for the effective range of the Level 1 and Level 2 tools to line up if they are to be administered in sequence. More recent research has shown ample evidence that many of the behaviors covered in the M-CHAT-R are present in children as young as 6-12 months (Miller et al., 2021; Hutman et al., 2011; Neimy et al., 2017; Winder et al., 2013). As such, I believe that administering the MASCOT-L1 at 12 months of age to catch any early strong presentations of ASD, and then again at the statistically validated 16 months of age, would be valuable, as many children could be given 4 additional months of valuable intervention. With this change, both levels of the tool could be administered in sequence with each other, without a confusing limbo period between when one can be administered and the other cannot.

Removal of Yes or No Scoring in MASCOT-L1

Currently, the M-CHAT-R employs a binary scoring system. Of the 20 questions, each yes or no response is coded as an at-risk or not-at-risk response. Upon completion of the survey, a score of 0-2 at-risk responses means the child is at low risk for ASD, a

score of 3-7 at-risk responses means the child is at moderate risk, and a score of 8-20 at-risk responses means that the child is at high risk. However, I believe that this scoring system is far too rigid for a disorder as heterogeneous as ASD. In situations where a child has exhibited a behavior only a few times, parents are told to answer no. Instead, I would incorporate a partial credit scoring system, which is a strength of the ADEC. For the MASCOT-L1, a behavior that has been observed but not with frequency would be scored as a half-point. As it does with the ADEC, this change would be helpful in screening for atypical presentations of ASD, where perhaps a broad range of behaviors are observed, but not frequently enough to be fully obvious to a parent with less education about the disorder.

Increased Sensitivity of MASCOT-L1

The balance between sensitivity and specificity is a concern with any diagnostic tool for any disorder, not just ASD. An over-sensitive tool would result in false positives, which has the negative effects of increasing parental stress (in the case of a Level 1 tool, more families would have their child flagged for moderate- or high-risk for ASD), longer wait times for diagnostic appointments, and putting children through examination sessions that may not be necessary. However, I argue that increasing the sensitivity of the Level 1 tool would have benefits that would far outweigh the drawbacks. An increased sensitivity would reduce the number of false negatives, resulting in fewer children who pass the screening tool as low-risk individuals but are eventually diagnosed with ASD. Recipients of a false negative result would likely miss out on critical developmental months where the lack of a diagnosis would result in no further intervention. As previously mentioned, the consequences of late diagnosis can have strong adverse effects on a child's development. I argue that a reduction in cases like

these is far more valuable than the lack of parental stress and wait times that result from a hyper-specific tool. In fact, it could be argued that giving more children a more detailed assessment could mitigate parental stress in the long-term: stress fueled by ambiguity and lack of understanding of a child's developmental barriers would be far less frequent if more children were administered the MASCOT-L2. Instead, more families would receive information as to their child's unique situation, and be referred to resources that could aid in their development. In making developmental screenings more common, this could also possibly serve a role in reducing stigma around labels such as ASD, and raise awareness around developmental delays as a whole.

Currently, the benchmark for a moderate-risk child would be a score of 3 or greater, and I would reduce this to 2.5 or greater, incorporating the aforementioned half-point scoring system. Analysis of the M-CHAT-R has shown that 1 in 127 children is flagged for moderate or greater risk (Robins et al., 2014), which is a stark difference from the CDC's 1 in 36 estimated prevalence (Centers for Disease Control and Prevention, 2023). Reducing the threshold for moderate risk would bring these figures closer together, allowing the tool to better reflect the actual prevalence of ASD in the population.

Risk-Based Priority System

In order to counterbalance some of the issues that would arise from increasing the MASCOT-L1's sensitivity, I also propose that this change would be accompanied by a system in which children who score as high-risk on the MASCOT-L1 would be able to receive their MASCOT-L2 assessment at a higher priority than children who score as moderate-risk. As a result, children with more clear-cut symptoms and needs would be able to obtain interventional resources such as therapy programs or diagnoses at a

similar, or even quicker rate than prior to the change in sensitivity. This priority system could take many forms, but simply reserving a set quantity of daily appointment slots strictly for children who were flagged as high-risk could allow for very quick assessments particularly for families with significant, pressing concerns about their child. With this system in place, the added concern of wait times that would typically come with an increase of sensitivity would be alleviated, at least for those displaying strong correlates in multiple domains of ASD.

Inclusion of Motor-Related Measures

As previously mentioned, motor-related criteria are not present in any of these diagnostic tools. However, adding some items to the MASCOT-L1 that mention motor deficits (for example, “Does your child struggle to maintain balance while sitting up or reaching for a toy?”), and including motor assessment during the diagnostic session would help address the heterogeneity of ASD, likely at very little cost. For those children with ASD who do not experience any motor issues at all, the inclusion of motor-related items would not affect their scoring. Instead, this would allow for infants that have just a couple of at-risk social or behavioral tendencies, but also some semblance of motor-related issues to get a second look that they would not otherwise receive. If the child showed concerns with motor issues on the MASCOT-L1, the MASCOT-L2 would include additional tasks that would examine this (based on the previous motor research (Iverson et al., 2019), this could include tasks such as reaching for toys and attempting to pull oneself up into a sitting position). Again, with the goal of reducing false negatives and a diagnosis at a later-than-ideal date, giving potentially atypical presentations of ASD or those with similar delays in these domains a closer look could have positive outcomes for a child’s development.

Limitations and Future Directions

Although I believe that this new framework would allow for a better screening and diagnostic system that would help reduce the average age of diagnosis, there are some limitations to this project. Primarily, there would certainly be an increase in false positives, at least at the screening level, which could create some adverse effects on parental stress, wait times, and other issues that could arise from a highly sensitive tool. However, this is a risk worth taking considering the benefits of monitoring more infants' development closely and could result in far better outcomes down the road, not just for the children themselves but for their families as well.

Additionally, the issue raised about the unreliability of parental self-report is a problem with the M-CHAT-R that the mentioned modifications would fail to address, and perhaps this issue would be exacerbated by the fact that the diagnostic session's administration would be directly impacted by the parental self-report.

There are certainly far more issues with the ASD diagnostic process, such as the fact that boys are diagnosed with ASD four times as frequently as girls (Centers for Disease Control and Prevention, 2023), and it is unclear whether or not this is a reflection of the actual epidemiology of the disorder, or if this a shortcoming of our tools in that they skew towards detecting ASD correlates that are common among boys, and fail to detect ASD correlates that are more common among girls. Additionally, the inequality of access to healthcare resources is another reason that some ASD diagnoses happen either later than the ideal window, or not at all. Unfortunately, these issues are beyond the scope of this paper.

Additionally, early detection is only the first step in the process of aiding children with ASD. After detection, the appropriate intervention must be put in place, but the

science of behavioral intervention of ASD is nowhere near complete. Some studies have claimed that it is often difficult to measure whether early intervention therapies are actually efficacious, and that our future gold-standard interventions are yet to be developed (Mcglade et al., 2023). Others claim that the potential adverse effects of early intervention have been outright ignored by researchers, and more work must be done to ensure that the measurable outcomes are truly positive for those living with ASD and their families (Witwer et al., 2022). Ensuring that intervention techniques are ethical and strongly evidenced-based is a key component of the research-practice cycle.

Future directions in research to help further this field could take many different forms. Continued research into very early correlates of ASD is an important domain to consider, as this research can be used to further enrich screening and diagnostic tools, as it does in the proposed MASCOT. Additional research into some of the discrepancies we see in diagnosis between AMAB and AFAB individuals is also of utmost importance, as current tools certainly could help to address this.

The continued pursuit of early detection of ASD and other developmental delays represents the importance of developmental psychology and healthcare for young children. Through advancements in screening tools such as the M-CHAT-R, ADEC, and ADOS, the push towards identifying concerns at a stage when intervention can truly make a difference is immensely valuable for individuals and families alike.

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Chapter 2: Addressing Motor Considerations in ASD Diagnosis

Addressing Motor Considerations in ASD Diagnosis

Roadmap

In the previous Chapter, I discuss many of the shortcomings of screening and diagnostic tools such as the M-CHAT-R and the ADOS. Although this paper covered a breadth of issues with these tools, this at times came as a trade-off with depth into specific issues. One topic that deserves to be examined in far more detail is the potential of the fine- and gross-motor differences oft-observed in young children with Autism Spectrum Disorder (ASD) to be utilized in the screening and diagnostic process.

As discussed in Chapter 1, despite the fact that ASD is often accompanied by differences in motor development, these considerations are not included in gold-standard measures, most likely due to their absence as a core feature of the disorder in the Diagnostic and Statistical Manual of Mental Disorders. Instead, these tools opt to specifically operationalize the definition seen in the DSM-5-TR, investigating impairment of communication and social interaction as well as restricted and repetitive patterns of behaviors.

In this chapter, I will examine the evidence supporting the connection between motor development differences and ASD, and I will interrogate whether or not this connection warrants their further inclusion within screening tools or even within the DSM-5-TR itself. I will examine in detail the specific types of motor trends we see in infants and young children both before and concurrent with an ASD diagnosis, as well as investigating the predictive value of motor concerns for future outcomes. Additionally, I will consider some hesitations that current researchers have about the inclusion of the motor domain, and whether or not their concerns have strong enough merit to outweigh the other side of the argument. Finally, I will identify what I believe to be the most

important motor-related concerns in the screening and diagnostic process, and will provide potential solutions to them if the literature suggests that they would improve outcomes for those with ASD and motor developmental concerns.

Motor Development and ASD

Although Autism Spectrum Disorder is characterized by social/communication difficulties and restrictive/repetitive behaviors, the connection between Autism Spectrum Disorder and atypical motor development has also long been noted. The exact frequency of this is somewhat in dispute: one recent study, commanding a sample size of 11,814, found that 86.9% of children with ASD were at risk for motor impairment (Bhat, 2020) whereas another study, with a sample size of 2,084, found that 35% of children with ASD met the criteria for significant motor impairment (Licari et al., 2019). The latter study noted that their figure was lower than other similar studies, but it remains that there is not full consensus on the prevalence. Although it is true that this feature is not universal to ASD, and thus not a core component in the same way other behaviors are, it is evident that differences in motor development are a very real experience for a significant portion of those with the disorder.

Additionally, this connection is consistent throughout all levels of the autism spectrum. Although research has shown that often those higher on the autism spectrum exhibit more impaired fine- and gross-motor scores compared to those lower on the spectrum, both groups still score differently than typically developing children (Kaur et al., 2018). Hence, the presence of motor differences is not dependent on the level of ASD.

The difference in motor development between those with ASD and typically developing individuals is not one that shrinks as the children get older, in fact, quite the

opposite. A meta-analysis of research surrounding this topic found that children with ASD exhibited increasingly poorer motor function compared to typically developing peers as their age increased. During early infancy (0-6 months), the difference between those two groups was small, but still significant, but by 19-24 months, far larger effect sizes were observed across numerous domains of motor function (Lim et al., 2021).

Despite this well-documented link between ASD and motor impairment, there is little that clinicians have done, both in diagnosis and treatment, to reflect the connection, at least on a scale commensurate with its prevalence. Although Licari et al. (2019) determined that 35% of children met the criteria for significant motor impairment, they also noted that motor difficulties were reported by diagnosing clinicians only 1% of the time. Even with existing evidence that motor-based interventions can have positive results on an individual's motor skill development (Ketcheson et al., 2017), they are not practiced anywhere near enough given the prevalence of motor difficulties within this population. With such a significant gap between the prevalence of motor concerns and the rate at which they are actually addressed, one must wonder if there is a true inefficiency in the screening/diagnosis/intervention process of ASD when it comes to this domain, and how this could be addressed. To do so, it is important to understand the different ways in which motor concerns can manifest in this population, and how early they occur in comparison to the core features of ASD itself.

Different Types of Motor Presentations

Fine vs. Gross Motor Skills

Although it is established that ASD and motor development are undoubtedly linked, the type of motor difficulties those with the disorder face are not all identical.

ASD seems to be correlated with differences in development of both gross-motor and fine-motor skills. Gross-motor skills include bilateral coordination, visuo-motor coordination (i.e. hand-eye coordination, ability to catch a ball), balance, and gait. Fine-motor skills include handwriting, drawing/copying skills, and manual dexterity (Bhat, 2020). The collective fine- and gross-motor skills encapsulate almost the entirety of all motor development. The fact that ASD can impact such a broad range of an individual's motor development is significant, and seems appropriate for a disorder often marked by its heterogeneity, as was discussed in Chapter 1.

Specific Motor Presentations in ASD

Many individuals with ASD exhibit motor stereotypies, which are a type of hyperkinetic movement disorder currently defined as involuntary, patterned, repetitive, continuous, coordinated, purposeless, and ritualistic movements, postures, or utterances, although their exact definition is somewhat under debate (Melo et al., 2019). Motor stereotypies actually are included in the DSM-5's definition of Autism Spectrum Disorder, as a subset of the features that make up the restricted/repetitive behaviors domain. Although motor stereotypies are not specific to those with ASD, they often suggest the possibility of the disorder. A meta-analysis of 37 different studies found that the prevalence of motor stereotypies in ASD ranged from 21.9% to 97.5%, with a median of 51.8% (Melo et al., 2019). This same meta-analysis also found that frequency of motor stereotypies was associated with younger age, suggesting that these behaviors manifest quite early on in development. Unlike other motor difficulties, the acceptance of motor stereotypies within the diagnostic definition of ASD is an interesting asymmetry to be explored. Stereotypies are nowhere near the only motor-related behaviors that are observed more frequently with ASD, thus their inclusion within diagnostic criteria can

only scratch the surface of the relationship between the disorder and the motor domain. Thus, it is important to discuss all of the behaviors that are not included under this umbrella.

Although ASD can impact fine- and gross-motor development in all kinds of ways, some presentations are certainly more common than others. One such example is the domain of balance, gait, and posture. Research has consistently shown that there is a significant discrepancy between young children with ASD and typically developing children when it comes to balancing tasks, including both static balance (standing on one leg) and dynamic balance (jumping, hopping) (Odeh et al., 2020). Differences in gait and postural control have been noted as well (Fulceri et al., 2019), with some speculation that a difference in domains like these could be identified in analysis of childhood home videos, affording clinicians more information to work with (Ozonoff et al., 2007). Because of how large-scale, and thus more noticeable, the movements associated with these domains are, it is likely that these are easier for parents and guardians to spot at home.

Just as motor stereotypies are more present at younger ages, there are numerous motor development milestones that also can differ between typically developing infants and those with ASD. Lavenne-Collot et al. (2019) found evidence that infants with the disorder crawl on their hands and knees significantly less frequently than their neurotypical counterparts. In this same study, children with ASD also showed a decreased frequency of sitting up without help and a later mean walking age in comparison. Because motor development milestones are such an important part of development that parents are often attuned to, these behaviors could also be noticed at home more frequently than more niche motor difficulties that infants may have.

However, this is also complicated by the fact that motor development delays can have numerous origins.

Another domain that those with ASD often significantly differ within is the development of visual motor integration (VMI) skills. Visual motor integration is defined as the ability to perceive and process information visually, and coordinate a motor response. VMI encompasses many skills, including hand-eye, fine-motor, and gross-motor coordination. A trio of studies done on the connection between VMI and ASD all came to the same conclusion that the disorder negatively impacts VMI skills (Carsone et al., 2021). Considering that milestones of VMI development begin early, at about 3 months of age, and standardized testing for VMI skill in children as young as two years old already exists in the form of the Beery-Buktenica Developmental Test of Visual Motor Integration (Carsone et al., 2021), this domain is certainly one of interest when the possibility of detecting ASD through motor development measures is considered.

Another research-backed discrepancy between neurotypical and ASD motor development is that of prospective versus reactive motor control. A study comparing high-risk (sibling of another child with ASD) infants to low-risk infants in a task of catching a rolling ball found an interesting discrepancy between the two groups. As the ball approached the infants, the low-risk group would, on average, reach predictively before the ball entered their reaching radius. In contrast, the high-risk group would typically only react once the ball had already entered a spot where they could reach it. This difference in ability in predictive motor control also seems to manifest early, as the infants in this study were just 10 months of age (Ekberg et al., 2015).

This collection of studies is a testament to not only the prevalence, but the breadth of motor difficulties for those with ASD. Additionally, the age at which many of these studies were conducted far precedes the average age of diagnosis (4 years, 3 months) established in Chapter 1. When it comes to the goal of early diagnosis, identifying the earliest behaviors and correlates for ASD is a crucial practice. The fact that the diagnostic criteria for ASD are grounded in behaviors that do not typically emerge until at least 2-3 years of age (Iverson et al., 2018) makes diagnosis anything but straightforward. Since these aforementioned motor developmental differences seem to occur even earlier than other concerning behaviors, an attempt to utilize them in screening practices could help reduce the age of diagnosis significantly, if it is determined that they are predictive enough of future core ASD symptoms, and specific enough to not strongly overlap with other developmental delays or disorders.

Motor Development and Interaction with Other Features of ASD

Not only do motor development difficulties often precede core ASD behavioral symptoms, but evidence suggests that they can also impact them in numerous ways. Especially early on in infancy, developmental milestones do not progress individually in isolation: they inform one another. Thus, a delay in certain motor development domains can affect how an infant develops in the social, language, and behavioral domains that are so important to current-day strategies for ASD diagnosis.

Language and Communication Development

The link between motor development and language milestones is a very well-researched connection that may have great implications for the inclusion of motor skills in ASD diagnosis. If one of the core features of Autism Spectrum Disorder, persistent difficulties in social communication (American Psychiatric Association,

2022), has a strong causal relationship with motor development, the argument for its utilization becomes far stronger.

Early motor milestones are crucial for infants, as they can shape how the children can interact with the environment that surrounds them. For example, when infants are able to sit up unsupported, they gain access to a new visual vantage point of their surroundings, and they acquire greater freedom of their hands to explore objects, as they no longer need them to prop themselves up. This freedom sets up countless new possibilities for interaction with people and objects to build a base of the perceptual and social information that is so important for communication (Libertus & Violi, 2016).

This idea that developmental milestones can cascade into other, seemingly unrelated domains is not just theory. Libertus and Violi (2016) conducted a longitudinal study concerning this topic and found that there was a significant relationship between the emergence of sitting skills around 3-5 months of age and subsequent language development at around 10-14 months of age. Research examining this relationship has continued to support the theory that early motor development can impact the severity of ASD-related traits. A study comparing six-month-old infants with autistic siblings (high risk) to infants with unremarkable genealogy found that not only does the former group spend less than half as much time in an unsupported seating position, but the act of sitting has significant implications for the development of vocalization, corroborating the findings of Libertus and Violi (Iverson et al., 2018). Although neither study was able to pinpoint the underlying mechanisms that may account for such a relationship, the research clearly shows that these domains are intertwined.

In addition to gross-motor developmental skills like sitting, even fine-motor skills with less obvious connections to environmental exploration seem to correlate with ASD

in infants. Choi et al. (2018) conducted a study comparing high risk infants later diagnosed with ASD to typically developing infants, and found that the fine motor skill growth between 6 and 24 months is significantly slower in infants later diagnosed with ASD, and this difference predicts expressive language skills at 3 years of age. Longitudinal studies of this nature are extremely important for answering questions surrounding the causality and specificity of motor difficulties and autism, and this one in particular brings further evidence to the table that development of seemingly unrelated skills can be connected under the hood.

The impact of motor development on language and communication continues well beyond early infancy. Another significant milestone, the ability to walk, has been found to predict language development skills, such as vocabulary. Naturally, the view of the world from a crawling position is far more limited than an upright one, thus walking infants often have greater access to objects and caregivers' faces. Once infants can walk, they can (and do) explore environments more efficiently and for longer (Kretch et al., 2014). This increase in locomotion has been found to correlate with increased communication, as initiation becomes far easier with the mobility of walking (Iverson et al., 2018). Infants with a high risk of ASD diagnosis have been found to begin walking later than low risk infants on average. Relative to low risk infants, who typically walked (defined as taking 3 unsupported steps) at 11.76 months, high risk infants did not begin walking until 13.14 months. This same group of infants differed significantly in their acquisition of new vocabulary. The low risk infants acquired 6.2 new words per month, whereas the high risk infants acquired just 1.85 (Iverson et al., 2018). In essence, delayed or less effective use of walking may reduce opportunities for exploration and interaction, which may disadvantage language learning. This relationship seems to be

especially exacerbated in infants at risk for ASD, implying that perhaps infants with delayed motor milestones should be monitored more closely for delays in other domains as well. However, motor delays are neither specific nor universal to just ASD, and thus cannot predict a diagnosis on their own.

Interestingly, not only does early motor delay correlate with language and communication development, but the opposite case has also been supported: young children with ASD with typically developed motor skills are more likely to eventually lose their diagnostic label. Taverna et al. (2021) found this to be the case with fine motor imitation skills. Subjects (aged 8-20) in this study were asked to imitate various complex hand and finger positions. Those with a current ASD diagnosis scored poorer on this task than both neurotypical participants and those with a former ASD diagnosis, the two of which had indistinguishable scores on the measure. The study supported the idea that motor skills tend to normalize along with social and communication skills, as well as restricted and repetitive behaviors. As such, when an ASD diagnosis is lost, motor skills also align with neurotypical results. The authors considered two theories for this: firstly, having strong motor skills may help heighten response to intervention, and secondly, those that lost an ASD diagnosis could be more likely to participate in more physical activities, thus improving in the motor domain. Future research would have to be done to determine the true directionality of this relationship, but the connection between motor skills and core ASD domains continues to be of note.

Restricted and Repetitive Behaviors

In addition to the language and communication domain, research has found evidence that restricted and repetitive behaviors, another core domain of ASD (American Psychiatric Association, 2022), are also correlated with motor development.

This may come as a little less of a surprise, considering the aforementioned connection between this core criterion and the motor stereotypies often observed in those with ASD (Melo et al., 2019). However, restricted and repetitive behaviors is an umbrella concept that expands beyond just motor stereotypies. Other behaviors such as rituals and routines, insistence on sameness, and restricted interests also fall under this category, each of which do not directly involve motor function in the same way. Despite this, some motor features still have been found to strongly correlate with restricted and repetitive behaviors. Notably, one study examined postural control in children with ASD, and found that those with greater postural sway also had more frequent restricted and repetitive behaviors (Randonovich et al., 2013). Another study (Fulceri et al., 2019) corroborated these findings. In this study, an association was detected between the motor functioning and restricted/repetitive behaviors of preschool-aged children with ASD. Additionally, this association predicted not just frequency, but also ASD severity. Essentially, poorer fine- and gross-motor function predicted a greater quantity of restricted and repetitive behaviors in the subjects.

However, not all studies were as successful at establishing this connection. Although Kadaras et al. (2021) found that those with ASD had greater levels of gross- and fine-motor delay, as well as more restricted and repetitive behaviors than the general population, it was not found that one variable impacted the other.

If motor development delays were able to successfully predict not only language and communication deficits, but restricted and repetitive behaviors just as well, it would seem obvious to pursue this avenue as a possibility for early diagnosis. The ability to predict both domains of ASD would allow for a powerful risk-assessment tool. However, the evidence seems to support the prediction of language and communication outcomes

far more effectively than restricted and repetitive behaviors. Whether this is a matter of research volume or whether the statistical trend is truly weaker is unclear.

Other Motor Predictive Features

Although perhaps not immediately relevant to early diagnosis, difficulties with fine- and gross-motor actions can result in other outcomes that could make the lives of those with ASD more challenging. One such domain, daily living skills, has a well-researched association with motor difficulties (MacDonald et al., 2013, Travers et al., 2022). In particular, the ability to dress oneself (e.g., buttoning, zipping clothes), bathing and showering, educational skills (e.g., calendar use), health management, and cleaning all shared a strong positive correlation with difficulties in conducting gross-motor actions (Travers et al., 2022). These findings continue to emphasize the ability of the motor domain to impact other types of skills. Additionally, these findings could have implications for clinical treatment of those with ASD, as the presence of motor difficulty in a client could spur clinicians to pursue some form of occupational therapy for the child.

In addition to daily living skills, other behaviors often associated with ASD are also exacerbated by motor difficulty. Social isolation and lack of physical activity, two behaviors that are often associated with ASD, also tend to become more pronounced when motor development is delayed (Ketcheson et al., 2017). Again, although this may have few implications for early diagnosis, the fact that delays in motor development seem so intertwined with behaviors associated with the core diagnostic domains of ASD seems to point to important implications for treatment and intervention after a diagnosis is determined.

ASD and Developmental Coordination Disorder

DCD's Potential to Address ASD Motor Issues

One of the complications of utilizing motor development delays in ASD diagnosis specifically is the fact that other disorders can also include similar traits. Although it may be established that these delays often precede ASD-related behaviors, they can also precede other neurodevelopmental disorders or simply exist on their own. One such disorder, Developmental Coordination Disorder (DCD), shares a lot of the same motor characteristics that have been discussed earlier in this chapter. DCD is characterized by motor coordination below expectations for the child's age, potential delays in early motor milestones like crawling and walking, and difficulties with fine- and/or gross-motor actions. For a DCD diagnosis, these motor deficits must interfere with academic achievement or daily living skills and must not relate to a medical condition or disease (American Psychiatric Association, 2022; Harris et al., 2015). However, there is a provision in the DSM-5 that allows for DCD and ASD to be co-diagnosed (Bhat, 2020).

Many of these requisite features align with the discussed motor-related traits that often precede ASD diagnosis. In particular, the lack of motor coordination (Odeh et al., 2020), delay of motor-related milestones (Lavenne-Collot et al., 2021; Libertus & Violi, 2016), and interruption of daily living skills (MacDonald et al., 2013, Travers et al., 2022) have all been observed in infants and children with ASD, as mentioned previously. As such, encouraging a co-occurring diagnosis of ASD and DCD as a way to acknowledge and document the motor difficulties of a child could help address the lack of services that this group tends to receive for that domain.

Barriers to DCD and ASD Co-Diagnosis

Despite this overwhelming similarity of features, there is a great discrepancy between children with ASD who could receive a co-occurring DCD diagnosis and those

who actually do. Bhat (2020) found that although 86.9% of children with ASD were reported to be at risk for DCD (defined as <10th percentile of motor performance), only 15.1% of that same group actually held a professionally diagnosed label of DCD or other motor delay. Bhat contended that this significant discrepancy between the presence of risk and the formal diagnosis of DCD is a testament to the under-representation of motor difficulties in children with ASD. Often, a lack of diagnosis makes pursuing therapy or intervention far more difficult: only 31% of the children in this study were receiving any physical therapy services. It seems that although the majority of those with ASD struggle with motor-related issues, only a small fraction actually receive services for it.

Although this discrepancy may seem like an obvious oversight, other research has contended that the fine- and gross-motor difficulties seen in those with ASD are fundamentally different from those seen in DCD, despite the apparent behavioral similarities. While general motor ability (balance, manual dexterity, catching, etc.) may be comparable between ASD and DCD, ASD may be distinguished by poorer performance on imitation of meaningful actions and more infrequent command gestures (Miller et al., 2023). If the actual presentation of features is truly different, it would not make sense to automatically roll the motor-related traits of ASD into a DCD diagnosis.

Even ignoring potential underlying neurological explanations for the lack of co-occurring diagnosis, some believe that to obtain a DCD label, its features cannot be explained by a separate neurodevelopmental diagnosis. However, this is unconvincing, since as mentioned before, there is a specific provision in the DSM-5 that allows for DCD and ASD co-diagnosis. Some clinicians struggle with the fact that this is such a

special case, and in many instances believe that this is not the best approach, and thus neglect to include it (Miller et al., 2023). Some features of DCD make it difficult to diagnose along with ASD: primarily, DCD is not recommended to be diagnosed until the age of 5, thus making it difficult to justify including in any very early diagnosis of ASD. This is often difficult, as infancy through early toddlerhood is an important therapeutic window for dealing with motor milestone delays (Miller et al., 2023). Families that receive an ASD diagnosis for their child before the age of 5 would have to return to a clinic for re-evaluation to obtain a DCD label, which adds much temporal and financial stress that could otherwise be avoided.

Additional Motor Features of ASD

Potential to Reduce Diagnostic Gender Gap

One feature of ASD mentioned in Chapter 1 is the fact that boys are far more likely than girls to receive a diagnosis. The root of this inequality is not fully clear: whether the true prevalence of ASD is higher in boys than girls, or whether our current screening tools are overlooking features of ASD in female children at a greater rate than with male children, due to the predominantly male samples often used during their research and development. Interestingly enough, our current test measures for motor skills do not seem to have this same difficulty detecting ASD-related motor problems in girls. In fact, there is a possibility for the inclusion of motor measures in ASD diagnosis to tip the scales back in the other direction. In a study examining the potential sex-based variations of presentation in ASD, Dillon et al. (2021) found that there was, in fact, some sex-based variability in the degree of motor-related parental observations. Parents of females reported consistent motor concerns, indicating motor delays as a first concern, endorsing more motor delays and indicating greater delays in motor milestones. In

contrast, parents of males more consistently reported language and communication delays as a first concern (Dillon et al., 2021). Considering that language and communication delays are a significant part of the ASD diagnostic process, and motor concerns are not, this finding could certainly explain at least a portion of the discrepancy between male and female diagnosis rates. In another study, girls with ASD, but not boys, presented altered motor anticipation when asked to reach for and drop a ball into a hole compared to their typically developing peers (Crippa et al., 2021). In this example, this specific skill seems to only be impacted when it comes to girls with ASD. If the motor domain were more involved in this process, it is possible that diagnosis would be more equal between the sexes.

Potential for Earlier Identification

One difficulty of ASD diagnosis discussed in Chapter 1 was the problem of symptom identification. Since the disorder is partially rooted in social/communicative behaviors, it is often difficult to identify them before a child is school-aged and introduced into a variety of new social situations. As such, many caregivers do not observe autistic traits in their children until then, leading to a later-than-ideal time of diagnosis. However, motor-related ASD correlates may not have this issue. Motor behaviors typically precede language development, one of the primary diagnostic indicators of ASD, boosting the potential for earlier diagnosis. As mentioned in Chapter 1, early diagnosis leads to early intervention and better outcomes. One exploratory study examining the timeline of motor delays found that children with ASD were delayed in 9 out of 11 early motor milestones when compared to neurotypical counterparts, including turning, sitting, reaching, crawling, and walking, among others. All of these early milestones occur before the age of 2, providing evidence for the early diagnostic

potential of monitoring motor challenges (Liu, 2012). This study, in addition to some of the other previously mentioned early motor features of ASD, seem to support the idea that by closely monitoring motor skills during infancy and early childhood, healthcare professionals can potentially identify red flags for ASD, allowing for earlier intervention and support to enhance developmental outcomes.

Arguments Against Motor Inclusion in Diagnosis

Given all of the reviewed research supporting the connection between motor difficulty/delay and Autism Spectrum Disorder, why aren't motor challenges used in screening and diagnosis? There are several potential reasons.

Neither Specific Nor Universal

The problem with redefining ASD criteria and using motor difficulty in ASD diagnosis is the fact that they are neither specific nor universal to the disorder. It is not specific in that numerous other diagnostic labels also include motor delays or difficulties in their own definitions, such as the aforementioned DCD and Intellectual Disabilities (Bishop et al., 2022). It is not universal in that although the majority of those with ASD seem to have motor struggles (Bhat, 2020), there is still a non-zero portion of people with ASD and without motor issues. Contrasting this with ASD's core criteria, both social/communication deficits and restricted/repetitive behaviors and interests are specific and universal to ASD (Bishop et al., 2022). With diagnostic tools having the primary goal of accuracy, it does not make sense to look for criteria that are not at the core of the disorder, which motor problems are not. As such, I believe that redefining core features is not an appropriate solution to the problem, as it would result in too many unintended adverse consequences.

Difficulties in Measuring Motor Impairment

There is also some hesitancy when it comes to the measurement of motor impairment in this population specifically. There is concern that other features of ASD, such as avoidance of non-preferred activities and social isolation, may result in poorer test scores on motor measures while not actually reflecting ability correctly: "individuals with Autism Spectrum Disorder may be uninterested in participating in tasks requiring complex coordination skills, such as ball sports, which will affect test performance and function but not reflect core motor competence" (Bishop et al., 2022, pp. 1374). However, this argument is complicated by the fact that motor impairments often precede the presentation of ASD-related traits chronologically in development. As a result, if motor-based test results are truly measuring disinterest in the task, the root cause seems less important than simply identifying a developmental atypicality at all. If early motor discrepancies are found in an infant, then further examination and observation as the child grows could help fill in those gaps over time, while still giving the child assistance with their developmental milestones. This point concerning the underlying reasoning behind motor evaluation seems to disagree more with the idea of co-diagnosing DCD and ASD than it does using motor tasks to help identify early developmental struggles.

How Should Motor Features of ASD be Utilized in Diagnosis/Treatment?

Identifying the Current Issues

Based on the examined literature, it seems evident that, despite the strong connection between the motor domain and ASD-related traits, it is not just under-utilized, but not utilized at all in the diagnostic process of the disorder. Despite some evidence-supported arguments that it should not be used, such as its lack of sensitivity and specificity to the disorder, or concerns that motor measures are less

effective at measuring true motor skill in this particular population, the sheer rate of motor-related concerns in the ASD community makes it difficult to ignore.

In addition, modifying procedures to include motor measures would potentially alleviate some current issues observed in the screening and diagnostic process. Firstly, the fact that very early motor milestones in infancy can predict future ASD diagnosis highlights the potential to reduce the average age of actually receiving the label. Secondly, the current gender disparity of diagnostic statistics for the disorder could be reduced by the inclusion of motor observation in the process; the fact that motor issues in the ASD community have been observed in a greater proportion of girls than boys points to this idea.

Even in the proportion of those with ASD who are properly and punctually diagnosed, often the lack of an official diagnostic label acknowledging their difficulties in the motor domain leads to a lack of support and/or intervention to help address them. Consequently, the fact that motor issues seem to exacerbate ASD-related behaviors makes this even more difficult to accept. Although the motor domain is often treated separately from language/communication and restricted/repetitive behaviors (or not at all), they are undoubtedly connected, as supported in the literature, thus a system for addressing motor concerns in this population could help reduce the degree of ASD symptoms as a downstream result.

Incorporating the motor domain into clinical practices would be able to help in multiple ways, depending on the true nature of the relationship between ASD and motor behaviors. At the simplest level, just recognizing the frequency of those that struggle with motor development would allow for easier intervention, and resulting improvements in motor skills. However, if motor delays are truly predictive of ASD as

the literature suggests, then early detection would allow for a mitigation of ASD-related symptoms, or at least an early referral into the ASD screening process.

Solution 1: Monitoring for ASD When Motor Milestones are Delayed

Although the argument for the connection between the motor domain and ASD-related behaviors is strong, it is still true that motor difficulties are not a core property of the disorder itself. As such, I agree that it doesn't make sense to diagnose an infant with ASD when there is a presence of motor delays, but an absence of the behaviors that actually make up the diagnostic label of Autism Spectrum Disorder. However, that does not mean that the connection should be outright ignored, especially since infancy and early childhood are such critical times for intervention, as established in Chapter 1.

One way to include the motor domain in ASD screening would be to educate caregivers and pediatric clinicians about the connection between motor milestone delays and Autism Spectrum Disorder, thus increasing the level of diligence of observation in the home and during clinical visits. If a parent of a child struggling with motor milestones had a better idea of the predictive value it has for autistic traits, scrutiny of at-home observation could increase, and consequently an early diagnosis of ASD, when warranted, would be more likely to occur. As mentioned previously, it is often difficult for caregivers to observe ASD-related behaviors in infants before they are thrust into larger social situations like school environments, so education about this connection would allow for high-risk infants to have a more watchful eye on their development in not just the motor domain, but concurrently in social and behavioral domains as well.

Because of how separately each developmental domain is currently treated, calling attention to the interplay between them is of paramount importance for early

identification of not just Autism Spectrum Disorder, but other neurodevelopmental disorders that may impact an infant's milestones. Some concrete programming that could be done to address this is to encourage clinicians to have a discussion and share resources with parents concerned about their child's motor development, calling attention to the fact that other social and communication deficits sometimes follow as a result. Caregivers would then be able to return home with a greater understanding of the breadth of behaviors to observe for, instead of simply fixating on the domain that they consider to be the 'problem'. As research in this field continues to unveil specific milestone delays that tend to correlate with future ASD diagnosis, such as crawling and sitting up, these can be included as particular behaviors to keep an eye on.

Solution 2: Motor Difficulty as an ASD Specifier

One commonly proposed modification to the diagnostic process is to include a specifier within the label of ASD. A specifier is an extension of a diagnosis that further clarifies the course, severity, or special feature of a disorder or illness (American Psychiatric Association, 2022). Autism Spectrum Disorder already has specifiers in the DSM-5-TR: intellectual impairment and language impairment. If this modification were to be enacted, clinicians would be able to diagnose a child with Autism Spectrum Disorder with an accompanying motor impairment, thus acknowledging the motor domain in official documentation. If this were the case, although it would not aid in earlier diagnosis, it would make it easier for those with ASD and motor impairments to receive holistic treatment that addresses each and every affected domain, not just the ones core to the disorder itself. Licari et al. (2022) support this revision to the DSM-5-TR definition, arguing that there is sufficient evidence for motor impairments to be considered as prevalent and functionally impactful as other domains that are already

considered specifiers. Considering research that has shown that only 1% of children at the time of their ASD diagnosis have their motor impairments clinically recognized (Licari et al., 2019), this change in diagnostic language would be able to pave the way for increased recognition, thus allowing children easier access to motor-related therapies and interventions.

Solution 3: Revisitation and Awareness of DCD

A potential alternative to adding a motor specifier to the Autism Spectrum Disorder diagnostic label itself would be to modify the Developmental Coordination Disorder language to be more inclusive to not only co-diagnosis with ASD, but also more open to allowing the diagnostic label to be given to a younger demographic, prior to age 5 as currently recommended (Licari et al., 2022). In addition to modification of its language, it is important for DCD to be more generally recognized, such that clinicians can effectively recognize and diagnose it. In a survey of 1297 parents, teachers and physicians, only 41% of pediatricians and 23% of general practitioners had knowledge of the condition (Wilson et al., 2012). Perhaps this is another explanation for the low levels of co-diagnosis of ASD and DCD, despite the evidence that the majority of those with ASD qualify for the label (Bhat, 2020). If the awareness of Developmental Coordination Disorder were to grow among clinicians, then children with early motor concerns would be able to pursue intervention. If there truly is a causal connection between motor deficits and the severity of ASD-related traits, then early intervention for the motor domain would potentially help improve outcomes of well-being for those that are later diagnosed with ASD.

Limitations and Future Directions

Although I believe that each of the proposed modifications to the Autism Spectrum Disorder diagnostic process would help improve outcomes for those with the disorder, especially with clinically significant motor difficulties, it is important to acknowledge that a modification in current practices may have downstream effects not immediately considered before implementation.

Parental Stress and Anxiety

Education surrounding children's developmental milestones is important, especially for the parents and/or caregivers responsible for caring for the child in the home, as they will have the longest and most consistent exposure to the child and their behavior. As such, understanding the type of development in motor, language, and social domains that is to be expected is important, as well as understanding that there is often a range in when milestones are achieved. One potential drawback of monitoring for ASD when just motor milestones are delayed is parental stress, when often a short delay is no cause for concern at all. Adding more to the mental plate of someone raising a young infant is not always preferable, but in this case I believe that the benefit of education about the connection between all of the developmental domains outweighs the risks that may befall the caregiver. However, this does not mean that it is to be ignored, and clinicians and future researchers should look for ways to package this information in such a way that stress and anxiety are mitigated, while the education itself still remains.

Modification of Diagnostic Criteria

Additionally, the modification of diagnostic criteria could also lead to unintended downstream effects on the impacted population. If these suggested modifications to the ASD and DCD labels were to be implemented, future research should be sure to monitor

the rates of diagnosis (and mis-diagnosis) and the developmental outcomes of those whose labels are impacted, to ensure that the changes are truly increasing access to treatment and not resulting in any negative unintended consequences.

Future Research in Motor-ASD Connection

Future research should continue to not only examine how motor development and ASD are related, but also attempt to answer why they are connected. Building an understanding of the causal links between features of motor development and neurodevelopmental outcomes is paramount for creating the most effective treatments possible. Currently, there is some skepticism around the true nature of the connection between the motor domain and ASD (Bishop et al., 2022), and researchers should investigate this to ensure that our current testing measures are truly monitoring what we believe they are.

Conclusions

Though there is a long history of research connecting motor impairments to Autism Spectrum Disorder in infants and young children, the motor domain has, for the most part, not been involved in the screening and diagnostic process. Contemporary studies have been able to connect early motor development traits to future ASD diagnosis, and have even suggested that there is a causal nature between the often-considered-separate developmental domains. As such, using early motor markers as a way of flagging high-risk infants for ASD could be a useful practice for helping early diagnosis and intervention, potentially reducing not just age-related, but gender-related disparities in the field. In addition, making modifications to language in the DSM-5-TR could help clinicians officially recognize children with motor-related delays and

difficulties in their documentation, leading to an increase in the currently-low rates of children with ASD receiving treatment for them.

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