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**SCIENTIFIC FORUM: COMMENTARY**

# How relevant is the framework being used with autism spectrum disorders today?

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**Abstract**
Camarata (2014) provides a comprehensive summary of the current state of the research on early identification and intervention for children with autism spectrum disorders (ASD). Extending on the foundations provided by Camarata, this commentary discusses the value of a diagnosis of ASD and questions whether there is sufficient evidence on which to base continuing calls for early identification and ASD-specific intervention. Gaps are highlighted in the evidence base, suggestions made about how to fill those gaps, and an alternative framework is proposed for achieving best outcomes for children with early developmental problems of the type seen in ASD and their families.

**Keywords:** *Autism, autistic disorder, autism spectrum disorder, children, diagnosis, early intervention, prognosis.*

## Diagnosis, natural history, and intervention—is the current framework working?

Camarata (2014) states that an important rationale behind early identification in autism spectrum disorders (ASD) is so children can receive intervention earlier and, thereby, experience more favourable outcomes. However, he also notes the lack of accuracy and robust evidence for early intervention, calling for action to find such evidence. To date, we have continued to justify the need for early identification based on key assumptions; (i) that we know what ASD is and can correctly identify it early, (ii) that we know the developmental trajectory of ASD when diagnosed early, and (iii) that early intervention improves the outcome, beyond what is expected by developmental trajectory, for all children we identify. However, the difficult truth is that we are not certain of any of these things. As the foundation and links of this framework (Figure 1) are uncertain, we suggest it is time to formulate a better one.


Figure 1. Model of the links between early identification, intervention, and outcome.

## Diagnostic categories

Categorization for a diagnosis can be valid and useful, but making a distinction between a "disorder" and "no disorder" invariably involves the application of a threshold that is arbitrary. To choose a threshold that is meaningful requires certain conditions to be met. You need a shared understanding of when something is a "problem" or "abnormal" as opposed to part of a range of "normal" or "typical". You need a clear understanding of how the diagnosis is different from other associated problems or comorbidities. You also need to know the natural history, taking into account change that is expected because of development, such that a diagnosis predicts outcome and is useful in terms of making decisions about interventions to be used. Next, a reliable and repeatable "test" is required that accurately differentiates disorder from no disorder. When the application of a threshold based on a "test" (clinical or biological) has a high sensitivity and specificity, then the diagnosis is accurate (high discriminative validity), and reliable and repeatable measurement is implied.

It is easy to appreciate that developing meaningful arbitrary thresholds that fulfil these requirements is more difficult when the "test" involves observational

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measures that are subject to interpretation and context, and when there is no natural point at which to turn a continuous variable (e.g., a type of behaviour seen in everyone but to varying degrees) into a category of "present" or "absent".

The value in developing categoric diagnoses is well known. A diagnosis can result in harm (such as false positives and negatives) even with accurate and meaningful diagnoses. The risk of doing harm by using diagnoses with poor accuracy and for diagnoses that do not have a strong link to intervention choice and outcome prediction is higher.

## Autism spectrum disorder as a diagnosis

Camarata (2014) points out that the ASD we see today has changed a great deal from Kanner's autism in the 1940s. Individuals currently diagnosed with ASD are often as different as they are alike, and ASD is far from a tightly defined group. No unifying cause has been identified for ASD, although it is thought that a combination of genetics and the environment play a role in its development (Happé, 2006). In the absence of predictive biological markers, ASD is diagnosed by a collection of observed behaviours. Although attempts have been made over the years to revise the Diagnostic Statistical Manual of Mental Disorders (DSM) to improve the specificity and sensitivity of the ASD criteria and capture the dimensions and extreme heterogeneity of ASD (Happé, 2011; Lord, 2012; Swedo, Baird, Cook, Happé, Harris, Kaufmann, et al., 2012), there is ongoing debate about whether recent iterations of DSM have achieved this. Most recently, ASD has been defined as a dyad of impairments, which include social communication difficulties and restricted interests and repetitive behaviours (American Psychiatric Association, 2013). Each of these behaviours form a continuum from normal variation to abnormal with no clear cut-offs. Diagnostic tools have been developed in an attempt to categorize children, but even gold standard assessment tools and experienced clinicians lack consistency in diagnosis for those children in the grey areas or who show milder symptoms (Bishop & Norbury, 2002). To date it has not been possible to establish a universal constellation of early emerging behaviours that can reliably predict ASD in very young children (Macari, Campbell, Gengoux, Saulnier, Klin, & Chawarska, 2012). It is also challenging to disentangle ASD from other developmental conditions and/or co-morbidities such as communication delays, attention deficit hyperactivity disorder, and developmental disabilities at a young age (Paul, Chawarska, & Volkmar, 2008; van der Meer, Oerlemans, van Steijn, Lappenschaar, de Sonneville, Buitelaar, et al., 2012; Veness, Prior, Bavin, Eadie, Cini, & Reilly, 2012; Wetherby, Woods, Allen, Cleary, Dickinson, & Lord, 2004). Importantly, each of these diagnoses will have very different trajectories, intervention needs, and outcomes.

## Developmental trajectory (natural history or prognosis)

When a family receives a diagnosis of ASD for their child, they frequently ask questions about their child's likely future: Will my child talk? Will they cope at school? Will they live independently? These questions are also common to parents of children with other communication-based neurodevelopmental disabilities such as specific language impairment or apraxia of speech. Trying to predict which late talkers will develop a true language impairment, or who will have severe and persistent speech sound disorder, are familiar dilemmas in speech-language pathology (Paul, 1996; Rescorla, 2011; Whitehurst & Fischel, 1994). Balancing early intervention for children who may grow out of a problem with the needs of older children who have stable and persistently disordered trajectories, all within constrained resources, has been an ongoing challenge across fields. One Australian study found children with communication disorders were being both over- and under-serviced (Skeat, Wake, Ukoumunne, Eadie, Bretherton, & Reilly, 2013). Other studies have indicated that children were under-served (McAllister, McCormack, McLeod, & Harrison, 2011; Ruggero, McCabe, Ballard, & Munro, 2012). Similarly, many children with ASD have needs that are not being met (Brookman-Frazee, Taylor, & Garland, 2010).

There is evidence that, while a diagnosis of autistic disorder is relatively stable, there is poor diagnostic stability for children who are high functioning and/or who present with fewer ASD traits such as in Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS) or Asperger's Syndrome (AS) as classified in DSM IV (Rondeau, Klein, Masse, Bodeau, Cohen, & Guilé, 2011; Woolfenden, Sarkozy, Ridley, & Williams, 2012). Successful intervention could be hypothesized to produce this diagnostic instability. However, it is worth noting that, in children with PDD-NOS or AS, diagnostic outcomes moved in both directions, that is, to a "diagnosis of AD" or "no longer meeting criteria for ASD" in some studies and only in the direction of "diagnosis of AD" in others. Perhaps surprisingly, a number of studies also found that intervention was not a predictor of diagnostic outcome (Berry, 2010; Chawarska, 2009; Eaves & Ho, 2004; Jonsdottir, Saemundsen, Asmundsdottir, Hjartardottir, Asgeirsdottir, Smaradottir, et al., 2007; Lord, Risi, DiLavore, Shulman, Thurm, & Pickles, 2006; Turner & Stone, 2007). While the distinction between predictors of diagnostic outcome and an effective intervention as assessed by a randomized controlled trial need to be emphasized here, it seems likely that the developmental trajectory is not accurately predicted

by diagnosis, especially when made at a young age, and that intervention is only one factor influencing outcome.

## Early intervention

It is common for children with a diagnosis of ASD to be directed to early intensive interventions in resource-rich locations, and many clinicians and families believe that this is the best way forward. A summary of the effectiveness of early intervention is beyond the scope of this paper, but a number of reviews exist (Howlin, Magiati, Charman, & MacLean, 2009; Reichow, Barton, Boyd, & Hume, 2012; Warren, McPheeters, Sathe, Foss-Feig, Glasser, & Veenstra-Vanderweele, 2011). Of these reviews there is general agreement that not all children benefit, and the types and magnitude of benefits seen vary from trial to trial. To date there are also few well-controlled studies for very young children. Parallel to the evidence base about intervention effectiveness there is developing evidence that the age at which a child receives intervention and the intensity of the intervention may not be as important as the type of intervention or the child's individual characteristics and developmental trajectories (Berry, 2010; Darrou, Pry, Pernon, Michelon, Aussilloux, & Baghdadli, 2010; Fernell, Hedvall, Westerlund, Höglund Carlsson, Eriksson, Barnevik Olsson, et al., 2011; Magiati, Moss, Charman, & Howlin, 2011). One recent study found that children who received intervention earlier did not have better outcomes than children who received intervention later. Rather, factors such as IQ, regression, and the presence of medical conditions such as epilepsy had a more significant impact on outcomes (Eriksson, Westerlund, Hedvall, Åmark, Gillberg, & Fernell, 2013). It seems likely that some individuals will benefit from early intensive intervention more than others and, importantly, we need to know this information when they receive their diagnosis.

## Evidence gaps

### *Clarifying trajectories*

Children who have been diagnosed with ASD have different developmental trajectories (Baghdadli, Assouline, Sonie, Pernon, Darrou, Michelon, et al., 2012; Dereu, Roeyers, Raymaekers, & Warreyn, 2012; Fein, Barton, Eigsti, Kelley, Naigles, Schultz, et al., 2013; Gotham, Pickles, & Lord, 2012; Howlin, Goode, Hutton, & Rutter, 2004; Woolfenden, Sarkozy, Ridley, & Williams, 2012). Some children are different from typically-developing children when they are toddlers, but "grow out" of these differences; some have differences that lead to "diffability" (a difference in ability that may not have a functional impact on an individual's potential and/or wellbeing), and some have differences that progress to disability (impaired function and participation). Evidence is needed about these developmental trajectories to allow development of important sub-groups of children who may have ASD behaviours at different ages.

The recent push for identification, and intervention as early as possible, means that some children who have an outcome of typical or "diffabled", rather than disabled, will receive a diagnosis of ASD. Children who have an outcome in the typical range or of diffability, without intervention, will not benefit from rigorous testing, labelling, and intensive intervention, and they may even suffer harm, not yet measured or apparent, as a result of this management approach. If we find a small prevalence of this sub-group of children, then the current push for early identification is justified. However, if there is a substantial prevalence of children whose trajectory is toward typical development or diffability then it is not.

Clarification of children who *do not respond well to existing intervention* is also needed. As Camarata (2014) notes, inadequate detail has been provided on poor responders in many of the intervention studies conducted to date. Further, many intervention studies have excluded children with co-morbidities such as epilepsy, ADHD, intellectual disability (all of which are not uncommon in ASD and are important for prognosis). Although there are often good methodological reasons for these exclusions, they ultimately prevent exploration of the various co-morbidities that may influence lack of intervention response. In turn, the study of highly selected samples prevents us from assessing optimal approaches for children with challenging co-morbidities. These children often have the highest needs, and evidence to guide management is urgently needed. A clear delineation of this "non-responder" sub-group is important to allow interventions to be tailored to the child and their family's needs and to allow intervention innovation to occur.

We also need to establish which children are *not going to improve without intervention*. We have an opportunity to change the developmental trajectory for children in this group, and are unlikely to cause them unnecessary harm. Much refinement and intervention innovation is also still needed for this group.

### *Individualized interventions*

At the present time when a child is diagnosed with ASD, the intervention the child receives may be based on a broad range of theoretical perspectives and approaches from developmentally-based social-pragmatic to discrete trial (Paul, 2008; Wetherby, Prizant, & Hutchinson, 1998). When there are no clear guidelines or evidence regarding which particular intervention is the most effective for which child or family, the intervention can be typically based on a number of factors including the preference, experience, and training of the clinician,

funding requirements, what is readily available in the community, effective marketing, and what parents can afford (Goin-Kochel, Myers, & Mackintosh, 2007; Green, Pituch, Itchon, Choi, O'Reilly, & Sigafoos, 2006). It has been found that there is a lag between what is proven to be effective in research to what is being used by community service providers (Stahmer, Collings, & Palinkas, 2005). Clearly there is a pressing need to examine which interventions work best for particular individuals and their families and to determine the optimal timing and dose of those interventions. Attempts are being made to develop implementation models that will facilitate community service providers in utilizing evidence-based interventions (e.g., Drahota, Aarons, & Stahmer, 2012). Other researchers are examining ideal methods for studying predictors of intervention response (Yoder & Compton, 2004) and individual predictors of intervention response (see Stahmer, Schreibman, & Cunningham, 2011 for a review). For example, Paul , Campbell, Gilbert and Tsiouri (2013) compared discrete trial and naturalistic language interventions for children with severe autism and minimal speech. Joint attention moderated response to both interventions, but the children with better receptive language pre-intervention did better with the naturalistic approach and the children with lower receptive language did better with the discrete trial intervention. Kasari, Paparella et al., (2008) and Kasari Gulsrud et al., (2012) found that children who had five or fewer expressive words prior to intervention benefitted more from a joint attention intervention compared to a symbolic play intervention. Yoder and Stone (2006) compared Picture Exchange Communication System (PECS) and Response Education Pre-linguistic Mileu teaching (RPMT). They concluded that PECS may be more effective for children at the pre-request stage and RPMT may be more effective for children with some ability to initiate joint attention. Other studies have investigated parent and environmental predictors of intervention response (Ben Itzchak & Zachor, 2011; Gabriels, Hill, Pierce, Rogers, & Wehner, 2001; Osborne, McHugh, Saunders, & Reed, 2008; Robbins, Dunlap, & Plienis, 1991). This direction is encouraging as it better takes into account the heterogeneity of ASD and allows for intervention to be tailored to needs and abilities, rather than based on a categorical diagnosis. It also reflects the direction of much healthcare research at the present time that advocates a "personalized" or "individualized" approach.

## Where to from here?

The mental health and developmental disability fields are starting to re-think their frameworks for diagnosis and intervention. There is a groundswell to develop solutions that bring together genetics, neural circuits, the environment, and phenotypes. Some studies have already taken the first step (Fischbach & Lord, 2010; Zwaigenbaum, Scherer, Szatmari, Fombonne, Bryson, Hyde, et al., 2011). Speech-language pathologists must engage in these exciting developments, and continue to challenge the current framework used in ASD.

Our proposed model (Figure 2) focuses on the child's development rather than asking whether the child has ASD. We assess the child's behaviours as individual dimensions rather than focusing efforts on an "all or none" diagnosis. We acknowledge that this is still a blunt instrument approach and that in the future genetic, epigenetic, neurotransmitter, neuronal developmental, environmental, and other yet unknown variables could shape intervention pathways. However, we believe that this model provides more flexibility for intervention innovation, less risk of harm, and greater opportunities for discovery of causal pathways. As such we propose that ASD should be put back into a developmental context and early intervention should be based on the child (their

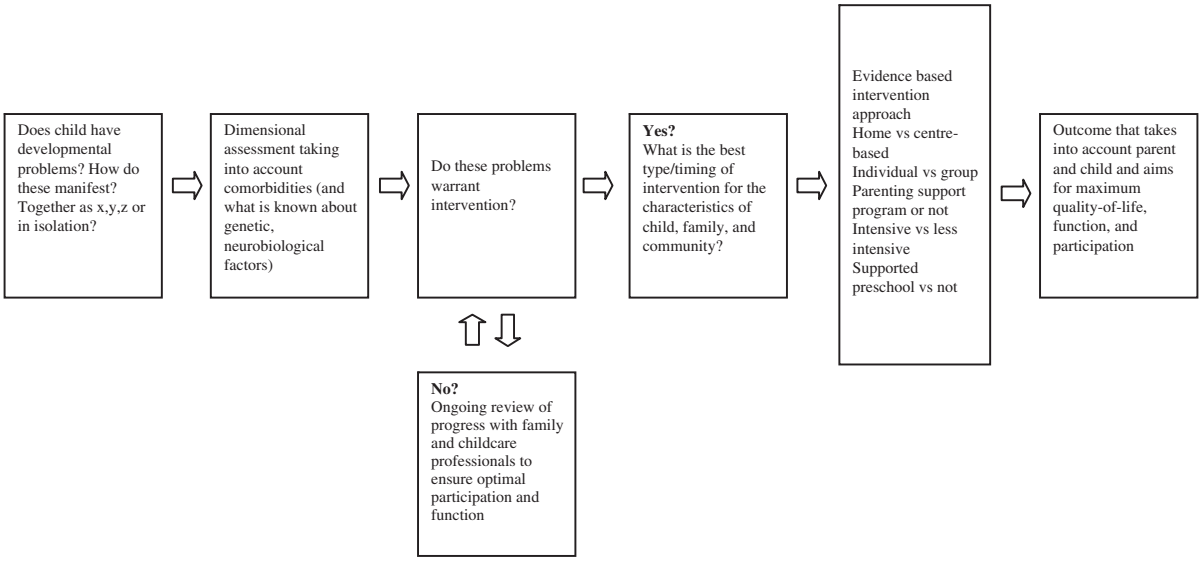


Figure 2. Alternative model for autism spectrum disorder (ASD) diagnosis and intervention.

family and community's) characteristics, degree of impairment, needs, functioning, capacities, and trajectories, not based on a specific diagnosis at one point in time.

We are not suggesting a watered-down, generic approach to intervention, but an approach that would allow us to consider a range of evidence-based intervention approaches, and the flexibility to use both targeted and universal service platforms and for any child to move between these depending on their abilities and developmental trajectory. In this model, the issues around trying to diagnose a child under two years of age with ASD are irrelevant. This model would also remove pressures in making a diagnosis so that a child does not miss out on interventions, supports and resources just because they don't meet an arbitrary cut-off. This approach would allow us to "do no harm" with a label that currently lacks accuracy and does not predict the outcome or direct intervention. Further, this approach is more consistent with the direction of research currently being undertaken internationally in mental disorders (e.g., Insel, Cuthbert, Garvey, Heinssen, Pine, Quinn, et al., 2010).

## Implications

It is important that clinicians be aware that the diagnosis of ASD is imperfect and in itself may not be useful in predicting an individual's intervention needs and outcomes, especially for young children. Children diagnosed with ASD are very heterogeneous and not all children respond positively to early intensive interventions. Importantly, we need to know more about the characteristics of the children (and their families) that respond differently to the various types and intensities of interventions. We also need to continue to be innovative in our thinking in order to meet the needs of children who do not respond well to the interventions developed thus far. Clinicians need to be willing to embrace the unknowns of ASD trajectories and work together to gather data about factors that describe and influence outcome at every clinical encounter. This includes dimensional assessments that take into account a child's comorbidities. Traditional, small, well conducted trials will not assist with this and may not be well suited to studying the inherent complexity of neurodevelopmental disabilities (Rosenbaum, 2010). Rather, population-based naturalistic studies may be required in order to understand how intervention is being translated in communities for a broad range of children. As our knowledge about ASD and its causes grows at an extraordinary rate it is important to place this new knowledge within the context of the frameworks currently used in ASD and to continuously ask questions about whether the two remain a good fit.

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