Early identification of autism spectrum disorder: Do we need a paradigm shift?

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Autism spectrum disorder (ASD) is a complex neurodevelopmental disorder characterised by deficits in social interaction and communication as well as restrictive/repetitive behaviours (American Psychiatric Association, 2013). The rate of occurrence of ASD is increasing steadily, with the rate progressing from 1 in 2500 children 40 years ago to 1 in 200 in the last decade to the current estimate of 1 in 68 children in 2014 (Frieden et al., 2014). At the same time, there are concerns that the current surveillance programmes are failing to identify children at developmental risk due to poor uptake, thereby missing opportunities for early intervention. Recognising early signs and symptoms of ASD is particularly challenging, and hence the American Academy of Paediatrics (AAP) has recommended that this be done within the broader framework of developmental surveillance alongside ASD-specific surveillance and screening algorithm (see Armstrong, 2008) during 18- and 24-month well-child visits. This is particularly relevant for the increasing number of children diagnosed with high-functioning autism (Frieden et al., 2014) where symptoms of speech and language delay may be less obvious and therefore the diagnosis may be easily missed. Furthermore, for some children, co-morbid behavioural or emotional difficulties might be the primary concern and mental health professionals need to give ASD due consideration while assessing young people with behavioural or emotional difficulties such as attention deficit hyperactivity disorder (ADHD) or social anxiety.

It is estimated that around 10% of parents will not raise any concerns in children identified as having ASD and around 50–70% of primary care professionals do not use standardised instruments for developmental screening at well-child visits although the diagnostic accuracy is poorer when clinicians rely solely on clinical judgement (Miller et al., 2011). Given the fact that early intervention is vital for improving outcomes by maximising the brain plasticity, no effort should be spared in offering this opportunity to every single child with a developmental profile suggestive of ASD regardless of the diagnostic outcomes. In this regard, routine use of the Edinburgh Depression Scale (EDS) for screening perinatal depression is a case in point. The use of EDS has helped primary care professionals to initiate a conversation about the new mother’s emotional status and identify those at risk of depression, although several criticisms have been made about its use (or misuse). These include the primary care professionals not using it at all or using it inappropriately as a diagnostic instrument when it is meant to be a screening tool, as well as issues around false-positive cases where women experiencing temporary unhappiness being given a diagnostic label and so on. However, the benefits outweigh these shortcomings in that it aids early identification of those with perinatal depression while also providing an opportunity for others who do not have clinically significant depression but yet whose low mood and problematic adjustments to motherhood deserve attention. There are also concerns that the threshold may be pushed for prescribing antidepressants in this group of women. In the case of ASD, however, such concerns are not valid. Instead, early identification coupled with a shift in the practitioner mindset from ‘watch and wait’ to early referral and intervention of those identified to be at developmental risk (regardless of diagnostic labels) can substantially improve outcomes.

A criticism could still be raised that at least some children and families may be put through unnecessary anxiety if further assessments do not yield an ASD diagnosis. This is where we need a paradigm shift in our approach to early identification. The same is true for breast cancer screening, but we accept that the risk of causing undue anxiety in some false-positive cases far outweigh the benefits. Both professionals and communities would need to accept such an approach in the case of developmental surveillance. With this in mind, parents should be encouraged to raise concerns just like we advocate self-examination of the breast coupled with community-based surveillance programmes similar to mammograms after a certain age. In this

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regard, universal developmental surveillance programmes (rather than just cross-sectional screening) recommend that age- and stage-specific assessments (at 6-monthly intervals starting from 6 months) be done during ‘well-baby’ checks with a particular focus on autism-related symptoms in the second year of life. A 2016 US Preventative Services Task force Recommendation Statement also suggests the importance of a parent–clinician partnership to elicit developmental concerns during routine primary care visits from 18 to 30 months as the critical first step followed by more specific assessments and early intervention as appropriate. Intervention may include enriched environments at home and at playgroups or early childhood centres for milder forms of developmental delay as well as intensive intervention for moderate to severe developmental delay and targeted intervention for ASD.

It is also to be borne in mind that ASD lies on a spectrum of conditions with significant differences in symptom constellations needing different interventions. In other words, ‘autism’ is ‘autisms’ which are both clinically and genetically heterogeneous. Although some of these genes are shared across different cognitive domains and clinical conditions including intellectual disability, ADHD and so on, through our attempts to classify behavioural symptoms in a meaningful way, we have created arbitrary divisions between clinical syndromes that do not neatly map to the underlying pathogenetic processes. Just like in hypertension there are different pathways such as ‘idiopathic’ cases and those ‘secondary’ to another medical or renal condition, some cases of ASD are secondary to another single gene disorder (syndromic ASD as in Tuberous Sclerosis), while in others, there are no identifiable aetiology. The latter are thought to be spread on a dimensional spectrum of traits across the general population with the suggestion that when it reaches a certain threshold due to a specific combination of risk and protective alleles, it manifests as broad autism phenotype (BAP) as may be seen in some first-degree relatives of patients, and when it crosses a second threshold, it manifests as clinically significant symptoms that we label as ASD (Eapen et al., 2013). Furthermore, the clinical presentation may change over time as a function of the change in developmental trajectory. Thus, the social and communication deficits would present differently in a toddler as compared to a school-aged child or an adolescent, and there is a need for multiple assessments to be conducted in a developmental context over time. Hence a framework of surveillance with ongoing monitoring is needed which can be best achieved by utilising the opportunistic immunisation contacts to enhance cost-efficiency. In this regard, it is worth noting that the Australian Government recently terminated the programme of the 4-year Healthy Kids Check by the general practitioners (GPs). However, this was a ‘one-off screen’ and is both clinically and theoretically significantly different to the ongoing surveillance programme as advocated by the AAP where children receive ongoing monitoring using valid and longitudinal surveillance tools from an early age (e.g. from 9 months for overall developmental progress and 18 months for autism-specific checks) rather than a one-off check (at the start of school at 4 years), followed by opportunities for further assessments as needed and assistance with accessing early intervention. Such a system has the potential to serve as a universal surveillance platform that is uniform across all states and territories that can translate to early intervention opportunities. This would link nicely with the Early Childhood Early Intervention (ECEI) approach within the National Disability Insurance Scheme which is designed to support intervention (regardless of diagnosis) at the earliest possible opportunity to achieve the best outcomes. This is also in keeping with the recently launched initiative of the Australian Government, namely, the ‘My Child’s eHealth Record’ which recommends ongoing developmental checks (using Parent Evaluation of Developmental Surveillance) at 6, 12, 18 months and 2, 3 and 4 years of age.

We also need to be mindful of the limitations of the ever-changing nosology and classificatory definitions in our diagnostic process so that they do not become barriers to accessing services as there are significant problems in the way our services are currently organised. We need to move away from the current model of service delivery based on diagnostic and classificatory boundaries with some services being only available to a certain clinical condition, to a more holistic approach whereby a child at developmental risk has access to services based on his or her functional needs and family/environmental context. There is increasing evidence that early intervention has the potential to alter adverse development towards a more neurotypical trajectory, thereby providing significant short- and long-term benefits to human capacity through increased school retention, reduced unemployment and welfare burden. However, we need to take on board the fact that just like all breast lumps are not cancers, not every child presenting with a developmental problem may reach a diagnostic threshold, and yet early identification and intervention offer the best outcome.

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