



ORIGINAL ARTICLE

Agreement between concern about autism spectrum disorder at the time of referral and diagnosis, and factors associated with agreement

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Aim: This study examined the level of agreement between referral reason and diagnostic outcome for pre-school aged children showing developmental delays and differences. In particular, the level of agreement when there was concern about autism spectrum disorder (ASD) was investigated.

Methods: Clinical records for children ($n = 677$) assessed for developmental concerns at a multidisciplinary assessment clinic in Sydney, Australia, were examined. Referral reason and diagnostic outcome were compared. Referral sources and factors including age, developmental level and language functioning, were explored for potential association with the consistency between referral reason and diagnosis.

Results: Approximately 30% of children referred for developmental assessment with ASD concern were not diagnosed with ASD. Agreement between referral reason and diagnosis was similar regardless of referral source and child age. Of children where diagnostic outcome was discordant with referral reason, 24% had delayed language. Variability in cognitive or developmental profiles was common (64%), regardless of diagnostic outcome.

Conclusions: Referral for ASD diagnostic assessments in the cases where ASD is not the primary cause for a child's difficulties increases pressure on assessment services, can delay access to ASD-specific intervention services for children who do have ASD, and can contribute to parent anxiety unnecessarily. The current evidence that a substantial minority of children were referred for ASD diagnostic assessments when ASD was not the primary cause for the difficulties emphasises the value of adopting a developmental differences framework, focusing on dimensions of behaviour and functioning, rather than an autism-specific service structure.

Key words: autism; autism spectrum disorder; developmental concern; diagnosis; referral reason.

What is already known on this topic

- 1 The prevalence of autism spectrum disorder (ASD) has increased over time as diagnostic criteria have broadened.
- 2 This is associated with greater public concern and professional awareness of ASD.
- 3 Increased ASD prevalence and concerns places greater pressure on ASD diagnostic and intervention services.

What this paper adds

- 1 A substantial minority of children with ASD concern during pre-school years do not meet ASD diagnostic criteria following assessment.
- 2 There is a tendency to be overly concerned about ASD in children with language delays.
- 3 A developmental differences framework, rather than autism-specific service structure, may provide a more meaningful approach to conceptualising and assessing developmental differences.

There is increasing public awareness and concern about autism spectrum disorders (ASDs).¹⁻³ This leads to early referral of children with atypical development or behaviour to assessment services seeking a diagnosis. Early referral can facilitate access to intervention programmes, providing opportunities to promote learning and participation when appropriate.^{1,4,5} A recent study,

however, reported that 39% of children referred to a regional ASD diagnostic clinic in the USA were not diagnosed with ASD following assessment.⁶ Thus, the heightened public awareness of ASD may also result in referrals to diagnostic centres for children who do not have ASD. An ASD diagnostic assessment for these children without ASD likely contributes to longer waiting times. This inevitably would delay diagnosis and access to ASD-specific intervention when indicated, and may also be associated with avoidable parental anxiety.

According to International Classification of Diseases, Tenth Revision (ICD-10)⁷ and Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5)⁸, the essential criteria for ASD are qualitative impairment in social interaction and

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communication, and the presence of restricted, repetitive and stereotyped patterns of behaviour, interests and activities. In the absence of biological markers for ASD, formulating a diagnosis requires comprehensive assessment of these areas, interpreted within the context of the child's medical and developmental history. Parent and teacher report is therefore important, together with direct child assessment of behaviour, language and developmental or cognitive level.^{9–12} Given the diagnostic complexity in ASD, inter-disciplinary team approaches are recommended across the UK, Europe, USA and Australia.^{9,10,12,13} The diagnostic team should at least comprise a paediatrician, psychologist and/or speech pathologist, with input from occupational therapists, neurologists and psychiatrists also recommended where possible.

Parramatta Early Childhood Assessment Team (PECAT) of Sydney Children Hospital Network, Australia, provides diagnostic and assessment services for pre-school children (aged 6 years and under) with delays in two or more unrelated domains of development. Concerns regarding ASD and/or other medical conditions may also be present. Consistent with practice guidelines, the diagnostic team consists of developmental paediatricians and psychologists. This study reviewed data for all children referred to PECAT between 2008 and 2010 to better understand the relationships among referral sources, child characteristics and assessment outcomes. During this period, referrals were received from parents, medical professionals (general practitioners (GPs), private and hospital paediatricians), nurses, allied health clinicians (occupational therapists, speech pathologists, physiotherapists and psychologists) and non-medical professionals from the community (e.g. child care centre and pre-school staff). Data for children referred with and without ASD concern were compared to examine the level of agreement between the referral concern and assessment outcome, and whether this varied according to referral source and/or child characteristics (e.g. child age, language functioning and cognitive or developmental ability).

Methods

Eligible children

Children ($n = 677$) referred and assessed at PECAT from 2008 to 2010 were identified from an existing clinical database. Study approval was granted by the Sydney Children Hospital Network Human Research Ethics Committee (LNR-2011-02-07) and was conducted in accordance with the provisions of the Declaration of Helsinki (2013).

Assessments

Developmental paediatricians and clinical psychologists completed child assessments and care giver interviews. A clinical nurse specialist or social worker was present as required. When ASD concern was noted by referrers or identified by PECAT clinicians, behavioural characteristics were elicited through parental interview and direct child observation. Reports from external informants were also collated, and the Childhood Autism Rating Scale (CARS)¹⁴ was completed. Clinical diagnoses were made based on all available information. Using DSM-IV criteria,¹⁵ the classifications were (i) autistic disorder, (ii) pervasive developmental disorder-not otherwise specified, (iii) Asperger's

syndrome or (iv) no ASD. For the purposes of this study, children with diagnoses (i)–(iii) were collectively categorised as ASD.

The Stanford-Binet Intelligence Scale-Fifth Edition (SBIS-5)¹⁶ and the Griffiths Mental Development Scales-Extended Revised (GMDS-ER)¹⁷ or 1996 revised version (if aged under 2 years; GMDS-1996)¹⁸ evaluated each child's cognitive or developmental level. Children were classified as within the *Average* range (SBIS-5 or GMDS/GMDS-ER classifications within the Borderline range or higher), or with *Mild, Moderate or Severe* delay (as per standard SBIS-5 or GMDS/GMDS-ER classifications). Children with substantial variability in subscale scores, preventing allocation of global category level, were classified as *Variable*. For the SBIS-5, a difference of greater than 10 points between verbal and non-verbal cognitive abilities, and/or non-uniform profile across subscale classifications (as described above), was defined as *Variable*. For the GMDS-ER or GMDS-1996, individuals with a disparity between domains greater than one category (e.g. *Average* fine motor development but *Moderate or Severe* delay in communication and language) were classified as *Variable*.

Data extraction

Data were extracted from the PECAT database. Data accuracy was verified by reviewing clinical records. Children were divided into two groups based on the referral reason: (i) ASD concern, including children with ASD features noted by referrers and children with a previous ASD diagnosis by a paediatrician or psychologist, and (ii) without ASD concern.

The data included age at date of referral and assessment; referral sources; referral reason (with/without ASD concern); diagnostic outcome; assessment tools used and test scores; and known associated conditions, including language delay, congenital abnormality, sensory (hearing or vision) impairment and behavioural disorders (attention-deficit hyperactivity disorder; oppositional defiant disorder).

Of the 677 children referred during the study period, 300 were referred and assessed in 2008, 186 in 2009 and 191 in 2010. The reduction in the number of assessments following 2008 was due to team relocation, involving re-housing and reduced availability of medical and psychological staff.

Statistical analysis

De-identified data were analysed using SAS version 9.3 (SAS Institute Inc., Cary, NC, USA). Continuous variables were described by mean and range, or median and inter-quartile range (IQR). Categorical variables were described by frequencies and percentages, and compared using chi-square tests for independent groups. McNemar's test was used to examine the level of agreement between the reason for referral and ASD diagnosis at assessment, and a kappa coefficient was calculated. There was no adjustment made for multiple statistical comparisons.

Results

Child characteristics

Of the 677 children reviewed by PECAT from 2008 to 2010, 35% were referred with ASD concern. This was similar across all 3 years, with 37, 31 and 34% referred with ASD concerns in 2008, 2009 and 2010, respectively.

There was evidence of an association between gender and ASD diagnosis ($P = 0.0006$), with males ($n = 151$) more commonly diagnosed with ASD than females ($n = 34$); male to female ratio approximately 4:1. Among the non-ASD group ($n = 492$), the gender ratio approximated 2:1 (M:F = 336:156).

Age at referral and diagnosis

The mean age at referral was 39 months (range 14–76 months). The majority of children were referred between 24 and 59 months, with a similar distribution of age of referral for children referred with and without ASD concerns (Fig. 1).

For children referred with ASD concern, the length of time between referral and ASD diagnosis ranged from 1 to 37 months (Md = 5.6, IQR = 3–7 months). ASD was diagnosed in 185 (27%) children aged between 19 and 126 months; median age of diagnosis was 44 months (IQR = 35–50 months). Age of assessment for children not diagnosed with ASD was similar (Md = 47 months, IQR = 35–54).

Agreement between referral reason and diagnosis

The referral reason and diagnosis agreed in 592 (87%) of cases (Table 1). The kappa statistic (proportion of agreement above what would be expected by chance) was 0.71. Of the 85 for whom referral reason disagreed with diagnosis, 67 (79%) were referred with ASD concerns but not diagnosed with ASD, whereas the remaining 18 (21%) were referred with other concerns but diagnosed with ASD ($P < 0.0001$, McNemar’s test).

There was no evidence that the agreement between referrer’s concern and the positive ASD diagnosis differed between age groups ($P = 0.11$; Table 2).

When consistency between referral reason and diagnostic outcome was considered for the full sample, the level of agreement was at least 80% for all referral sources ($P = 0.42$). Table 3 displays the agreement between reason for referral and diagnostic outcome for each referral source, with the sample differentiated according to referral reason. Of the 234 children referred with concern about ASD, the percentage of cases with confirmed diagnosis was similar across all referral sources.

There was no evidence that the level of agreement between referral reason and diagnosis differed according to the developmental level of the child ($P = 0.20$). As displayed in Table 4, 64% ($n = 435$) of children referred to PECAT had variation of ability for different aspects of their intelligence (i.e. intelligence quotient (IQ)) or developmental (i.e. developmental quotient (DQ)) profiles. Of these, 39% ($n = 171$) were referred with ASD concern, with 30% ($n = 129$) meeting diagnostic criteria for ASD.

The number and proportion of children with a language delay, known congenital condition, sensory (hearing or vision) impairment or attention-deficit hyperactivity disorder/oppositional defiant disorder are shown in Table 5, according to referral reason and diagnosis.

The level of agreement between referral reason and diagnosis differed according to the presence of associated conditions ($P < 0.0001$). Of 324 children with no associated conditions, agreement between referral reason and diagnostic outcome was 89% ($n = 288$). Agreement between referral reason and diagnostic outcome was 75% ($n = 126$) in children with language delay ($n = 168$). Of the 42 children with language delay where the referral disagreed with diagnosis, 40 (95%) were referred with ASD concern but did not meet diagnostic criteria, while only two children (5%) referred without ASD concern were diagnosed with ASD following assessment.

Discussion

In this study, a substantial minority (approximately 30%) of children referred for developmental assessment with ASD concern did not meet ASD diagnostic criteria, with similar patterns of agreement between referral reason and diagnosis regardless of child age and referral source. Only a small proportion (4%) of children referred without any ASD concerns were diagnosed with ASD. This indicates that, when developmental concerns were identified and further assessment was sought, few children meeting ASD criteria had been missed by referrers. Children referred with concern about ASD who did not meet diagnostic criteria frequently had delayed language development. Variability in cognitive or developmental profiles according to DQ or IQ assessments

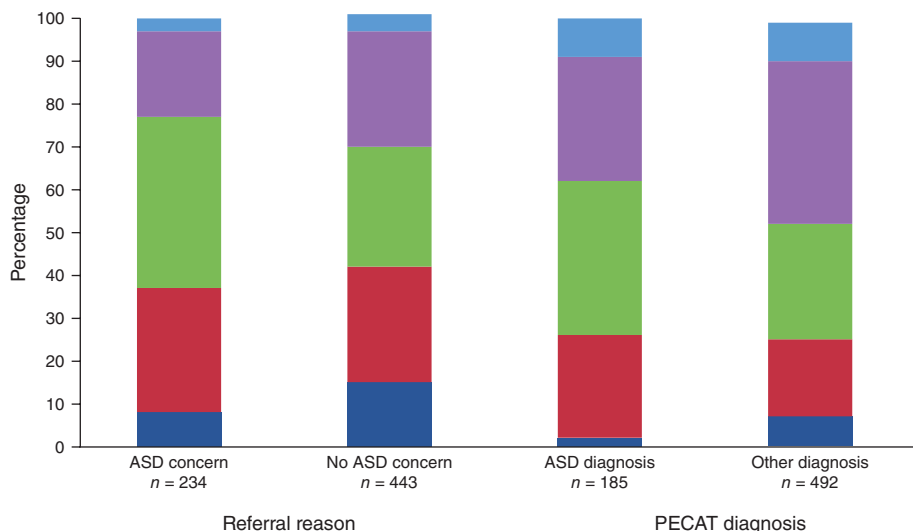


Fig. 1 Age distribution at time of referral and diagnosis. (■), ≥ 60 months; (■), 48–59 months; (■), 36–47 months; (■), 24–35 months; (■), < 24 months. ASD, autism spectrum disorder; PECAT, Parramatta Early Childhood Assessment Team.

Table 1 Agreement between reason for referral and final diagnosis

Referral reason	ASD diagnosis <i>n</i>	Other diagnosis <i>n</i>	Total <i>n</i>
ASD referral	167	67	234
Other referral reason	18	425	443
Total	185	492	677

ASD, autism spectrum disorder.

was common, with no evidence of difference between children diagnosed with ASD or other disabilities.

Recent estimates of the Australian ASD prevalence approximate 2%.¹⁹ This has increased over time in association with changing diagnostic criteria and inclusion of less severe differences for each criterion. The resultant wide variability in the clinical presentation of ASD can add to the complexity of diagnostic decision-making. Diagnostic complexity and growing evidence

supporting benefits of early diagnosis and intervention likely steers clinicians towards referring children for more comprehensive assessment when uncertain. In Australia, definitive ASD diagnosis prior to age six is a pre-requisite for accessing national funding. This has created a potential conflict for clinicians, where a diagnosis may be made to support a child's access to early intervention services, despite there being diagnostic uncertainty.²⁰ Greater community awareness also facilitates earlier detection and referral for ASD diagnostic assessments. In doing so, however, this increases pressure on diagnostic and intervention services.

Increasingly, there is a focus on ASD surveillance and screening from early childhood ages.^{21–23} This promotes early referral for diagnostic assessment when ASD is suspected, and timely access to intervention services when appropriate. However, there is debate about whether there is sufficient evidence for this approach, with potential negative impacts acknowledged for families in which children were falsely identified.^{24,25} Our results showed a substantial minority of children referred for ASD

Table 2 Number and percentage with agreement between referral reason and diagnosis by age

Age	Referral: ASD concern <i>n</i> = 234				Referral: no ASD concern <i>n</i> = 443			
	ASD diagnosis <i>n</i> = 167		Other diagnosis <i>n</i> = 67		ASD diagnosis <i>n</i> = 18		Other diagnosis <i>n</i> = 425	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
<24 months	3	75	1	25	1	3	35	97
24–35 months	36	72	14	28	8	10	76	90
36–47 months	63	78	18	22	4	3	117	97
48–59 months	51	68	24	32	3	2	162	98
≥60 months	14	58	10	42	2	5	35	95

ASD, autism spectrum disorder.

Table 3 Referral sources and PECAT diagnosis for children with and without ASD concern

Referral sources	Referral: ASD concern <i>n</i> = 234				Referral: no ASD concern <i>n</i> = 443			
	ASD diagnosis <i>n</i> = 167		Other diagnosis <i>n</i> = 67		ASD diagnosis <i>n</i> = 18		Other diagnosis <i>n</i> = 425	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
Paediatrician	77	70	33	30	6	5	121	95
ADHC	7	100	0	0	2	6	29	94
Psychologist	3	100	0	0	0	0	0	0
Carers	25	68	12	32	4	4	86	96
Community RN	6	100	0	0	2	9	21	91
DOCS	3	75	1	25	0	0	16	100
EI therapy team	29	71	12	29	2	2	85	98
GPs	2	50	2	50	1	6	15	94
Pre-school staff	15	68	7	32	1	2	52	98

ADHC, State Department of Ageing, Disability and Home Care providing therapy and case work services; ASD, autism spectrum disorder; DOCS, State Department of Community Services; EI therapy team, Early Intervention services providing physiotherapy, occupation and speech therapies; GPs, general practitioners; PECAT, Parramatta Early Childhood Assessment Team; RN, registered nurses.

Table 4 Level of agreement between referral concern, PECAT diagnosis and child intellectual/developmental level

Intellectual/developmental level	Referral: ASD concern <i>n</i> = 234				Referral: no ASD concern <i>n</i> = 443			
	ASD diagnosis <i>n</i> = 167		Other diagnosis <i>n</i> = 67		ASD diagnosis <i>n</i> = 18		Other diagnosis <i>n</i> = 425	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
Average	5	36	9	64	1	2	42	98
Mild	15	62	9	38	1	2	51	98
Moderate	5	45	6	55	1	4	25	94
Severe	6	100	0	0	3	5	53	95
Variable profile	129	75	42	25	12	5	252	95
Not assessed	7	87	1	13	0	0	2	100

ASD, autism spectrum disorder; PECAT, Parramatta Early Childhood Assessment Team.

Table 5 Associated condition by reason for referral and diagnosis

Associated condition	Referral: ASD concern <i>n</i> = 234				Referral: no ASD concern <i>n</i> = 443			
	ASD diagnosis <i>n</i> = 167		Other diagnosis <i>n</i> = 67		ASD diagnosis <i>n</i> = 18		Other diagnosis <i>n</i> = 425	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
None	132	79	23	34	13	72	156	37
Language delay	25	15	40	60	2	11	101	24
Congenital	9	5	2	3	2	11	165	39
Special senses	1	1	0	0	1	6	3	1
ADHD/ODD	0	0	2	3	0	0	0	0

Language delay: Specific language and communication delay according to Griffith's Mental Developmental Scales, as defined by score <70 on Scale C only (Language and Communication scale). Special senses: Hearing/sight impaired under one associated condition. Congenital: All known syndromes, congenital abnormality, epilepsy and prenatal insults. ADHD/ODD, attention-deficit hyperactive disorder/oppositional defiant disorder; ASD, autism spectrum disorder.

diagnostic assessment did not meet diagnostic criteria. This is consistent with recent research in the USA, where 39% of referrals for ASD diagnostic assessments did not have ASD.⁶ This places greater burden on already pressed diagnostic services, potentially delaying access to assessments and intervention for children who do have ASD. ASD diagnostic assessment when ASD is not the underlying cause of the child's difficulties can also increase parental anxiety. It is therefore important to carefully consider factors that may contribute to inappropriate referrals for ASD diagnostic assessments.

We have found that language delay, and not the level of global or cognitive development, may be an important clinical marker contributing to referral for ASD assessment in cases where ASD diagnostic criteria are not subsequently met. This suggests that referrers are overly concerned about ASD in children with language delays. While delayed language development has commonly been identified as one of the earliest markers raising concerns about ASD,^{26–28} such difficulties are not specific to ASD,²⁹ and 60–80% of children with delayed expressive language as toddlers 'catch up' by pre-school age.^{30–32} Thus, in cases

where language delay is a primary referral concern, seeking expert speech and language assistance early, and adopting a 'watch-and-wait' approach when language differences are mild, may be justified.

This study raises broader considerations regarding service configuration of developmental assessment services. It is not unusual for assessment and intervention services to be separate, and only loosely connected to primary care services. Increasingly, autism-specific services are also developing. Evidence that approximately 30% of ASD referrals do not meet ASD diagnostic criteria suggests that these approaches may be problematic. There is potential to focus on ASD-specific behaviours, or lack thereof, rather than interpreting functioning within a broader developmental differences framework. There is also the risk of assessment findings not being optimally linked to delivery of care and intervention.³³ Emerging evidence supports the importance of service structure emphasising links between multidisciplinary assessments and secondary care, with parents valuing information regarding their child's strengths and weakness gained from multidisciplinary assessments, but finding recommendations from sole paediatricians more useful.³⁴

This study offered the unique opportunity to compare the consistency between referral reason and diagnostic outcome across a number of referral sources. To the author's knowledge, this is the first study to explore Australian clinical data in this way. Evidence that the level of agreement was comparable between paediatricians and other referral sources, including non-professionals, was somewhat unexpected. This is not taken to suggest that paediatricians were underperforming in this regard; as paediatricians are able to diagnose ASD independently, without necessarily needing input from external assessment agencies, this may reflect a tendency for paediatricians to refer in cases where complexity and diagnostic uncertainty is greatest.

Limitations

Findings of this study relate to children aged 6 years and under, and cannot be extrapolated to older age groups. The data were gathered from a single diagnostic service only; the level of agreement between referral reason and diagnosis may differ to that found in other clinics. Data were limited to information available at the time of referral and initial assessment only. Given recent reports of lack of stability in ASD diagnosis over time,³⁵ repeat assessment during later childhood years would have been informative.

Conclusion

Current evidence that a substantial minority of children suspected of having ASD did not receive a diagnosis indicates that attempts to differentiate between diagnoses is challenging for pre-schoolers prior to comprehensive assessment. Referral for ASD diagnostic assessments in cases where this is not the underlying cause for the presenting difficulties places increasing pressure on services, results in longer wait times, and creates barriers to accessing intervention services in instances where a diagnosis is required for entry. Similarly, referral to autism-specific early intervention services prior to comprehensive assessment would have over-burdened those services, and potentially prevented access to them for children with ASD. Moving forward, a developmental differences approach where children are referred for comprehensive assessment because of functional or behavioural concerns could prevent unnecessary anxiety for families and reduce waiting times for assessments, if in turn that approach reduced the need for ASD-specific assessments for some children.

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The Donnybrook Hotel by Luther Miles (age 6) from Operation Art 2016