

# ASD and DLD diagnoses after 6 years old

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Delegates have access to a wide variety of comprehensive guidance material. If Delegates require further information on access or planning matters they are to call the TAPS line for advice. The Research Team are unable to ensure that the information listed below provides an accurate & up to date snapshot of these matters

**Research questions:**

1. What are the types of later life developmental disorder diagnoses?
2. What is the incidence of diagnoses for Autism Spectrum Disorder (ASD) and Developmental Language Disorder (DLD) that occur after the age of 6?
3. What is the incidence of diagnosis for ASD for age groups:
  - 0-6
  - 7-15
  - 16 and above?
4. What is the impact of a later diagnosis on the functional capacity and severity of symptoms of people diagnosed with ASD or developmental delay?
5. Are there triggers or acute events that precipitate diagnoses?
6. What is the impact of the resolution of an acute event on functional capacity regardless of diagnosis?
7. Are there therapies / treatments / protocols designed for people with later in life diagnoses?
8. What is the impact on prevalence of changes to ASD criteria between DSM-IV and DSM-5?

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## 2. Summary

Researchers continue to improve early identification methods targeting developmental disorders. This can reduce the waiting time for children to be diagnosed and for intervention to begin. In some cases, children do not receive an accurate diagnosis until later childhood or adolescence. Some are not diagnosed until adulthood. This paper focusses on the incidence and impact of diagnosing Autism Spectrum Disorder (ASD) and Developmental Language Disorder (DLD) after the age of 6.

There is limited information directly answering the research questions for an Australian context. I have gathered information relevant to the research questions which may approximate answers.

Issues related to overall prevalence of ASD have been investigated in another TAB research paper, [RES 222 ASD diagnoses](#).

### Types of later life developmental disorder diagnoses

Neurodevelopmental disorders (NDD) are a subset of developmental disorders defined by the Diagnostic and Statistical Manual of Mental Disorders, 5<sup>th</sup> edition (DSM-5). They are one of the most common classifications of childhood diagnoses and clinicians aim to diagnose the child as early as possible. Personal, clinical, social and environmental factors can delay diagnosis.

### Incidence of later life diagnoses

No reliable and comprehensive estimates were found for incidence of later diagnoses of ASD and DLD nationally or across all age groups. There is evidence that later diagnoses of DLD are common in people with a history of involvement with youth justice and child protection. Internationally, the latest systematic review finds the mean age for first diagnosis of ASD is 60.48 months (5 years). However, the best data for Australia suggests mean age of first diagnosis is 6 years for children diagnosed before the age of 13. This is likely to be considerably higher if older age groups are incorporated into the estimate.

### **Events leading to later life diagnoses**

There is evidence suggesting personal, social and environmental factors can predict whether someone will receive a later diagnosis of ASD. However, there is very little evidence describing events that precipitate a diagnosis. One study suggests adults choose to begin the assessment process due to encouragement by parents or spouses, difficulties with social interaction or mental health issues.

### **Outcomes for people with later life diagnoses**

There is evidence establishing the effectiveness of early intervention for people with ASD. There is less evidence establishing the adverse outcomes for people with later diagnoses though existing evidence does support the correlation of reduced functional capacity and increased comorbid conditions in people with later diagnoses. People with missed diagnoses of DLD are overrepresented in the youth justice and child protection systems.

### **Supports for people with later life diagnoses**

All interventions should be age-appropriate and targeted at the person's developmental stage. For older people this may mean interventions targeted at achieving life-stage outcomes such as employment and independent living. For people with ASD this may also mean accounting for the likelihood of comorbid conditions.

### **Prevalence of ASD after DSM-5**

Refer to [RES 222 ASD diagnoses](#) for further information. The restriction of DSM-5 diagnostic criteria for ASD has contributed to a reduction in the number of ASD diagnoses even as the prevalence of ASD continues to rise. The rise in prevalence should be attributed to factors other than the change in diagnostic criteria in the DSM-5.

## **3. Diagnosing developmental disabilities later in life**

The DSM-5 defines a group of NDDs which begin to manifest early in life and usually before the child enters school. NDDs can be global (affecting general intelligence or social skills) or specific (affecting specific aspects of learning or control of executive function) (DSM-5, 2013, p.31). They include intellectual disabilities, communication disorders, ASD, attention deficit hyperactivity disorder, specific learning disorder and motor disorders (including movement and coordination disorders and tic disorders) (p.xiv-xv).

Diagnosis of NDD is one of the most common types of diagnoses among children. Among NDDs the most common are learning disorders (8%), DLD (7%), ASD (2%) and ADHD (2%) (Micai et al, 2020, p.183). Behavioural signs are often observable within the first year of a child's life and some indications are known prior to the child's birth. For example, if the child has a sibling or other family member with a diagnosis of NDD it increases the risk that they will also have an NDD (Micai, 2020).

There are several factors that may delay a diagnosis. For example, it may be hard to determine if a child has social communication deficits until they are in situations which demand more sophisticated social behaviour. NDDs are often co-occurring, which introduces the risk of one diagnosis overshadowing other potential conditions and leading to later or missed diagnoses. Diagnoses can be a lengthy and costly process, which may delay the diagnosis itself or discourage some parents from beginning the process at all (Valentine et al, 2020; Micai et al, 2020).

## 4. Developmental Language Disorder

Many children show significantly slowed language development before the age of three. However, most of these children catch up to their peers after the age of three allowing them to perform within normal limits on linguistic tasks. Children who do not catch up may be diagnosed with DLD (Sansavini et al, 2021, p.2). DLD is the recommended label for language disorders that are not associated with a specific cause (e.g. autism, down syndrome). The term Specific language impairment (SLI) has also been used but the applications differ slightly (McGregor, 2020, pp.39-40). Prevalence of DLD among children is roughly 7% (Walker & Haddock, 2020, p.2; Ebbels et al, 2016, p.2). A report from the Deeble Institute of Health Policy states that prevalence among children in Australia may be as high as 17%, with higher rates in children from disadvantaged backgrounds (Walker & Haddock, 2020, p.2). This estimate is unreliable. The authors admit the estimate is based on minimal data and they do not offer an age range for the estimate.

I could not locate information on incidence of diagnoses by age. However, a 2021 review of systematic reviews suggests that the optimal screening time for DLD is between 2 and 3 years, with a diagnosis expected around 4 years (Sansavini et al, 2021, p.2). However, evidence is mixed with earlier screening increasing risk of false positives and later screening increasing risk of negative consequences for the child (p.20).

There is some evidence that DLD is often missed entirely or misdiagnosed in childhood (McGregor et al, 2020, p.40). A 2013 Australian study of 1607 children found only 45% of children with communication problems received any help before the age of 5 and only 33% received speech therapy (Skeat et al, 2014, p.219; Walker & Haddock, 2020, p.4). This does not differentiate communication problems from DLD specifically and so we can't straightforwardly conclude that most children with DLD are undiagnosed. It should also be noted that this study was prior to NDIS making early intervention available to more families.

A more recent Australian study of young people leaving 'out of home' care found that mean language scores were 2 standard deviations below the average. This level of deficit is often used as an indication of DLD. Despite this only a single participant in the study had a diagnosis relating to language difficulties (Snow et al, 2020, p.155). In another study of 44 young people leaving 'out of home' care between 16 and 26 years of age, Clegg et al found that over 60% met criteria for DLD and yet none had a diagnosis (Clegg et al, 2021, p.2). Results are similar in a youth justice setting (Snow et al, 2020, p.153; Clegg et al, 2021, p.2) with Winstanley et al finding 60% of their sample of youth offenders having met criteria for DLD despite no previous diagnoses (2021, p.399). These findings point to a high rate of unidentified DLD in young people with involvement of child protection or justice and is consistent with findings of previous studies that children from disadvantaged backgrounds are more likely to experience language difficulties (Walker & Haddock, 2020). Because the sample sizes of the studies were small and the populations unrepresentative, it is not possible to use them to reliably estimate prevalence of undiagnosed adolescents or young adults.

According to Walker and Haddock, research into the long-term effects of language impairment in an Australian context is limited. However:

International longitudinal studies have found that children with language disorders who do not receive intervention achieve lower levels of education and are subsequently at higher risk of lower wages and reliance on welfare and of higher levels of redundancy, under-employment and workplace conflict (2020, p.3).

As of 2016, most studies into the effectiveness of generalised intervention for symptoms of DLD in school-age children (i.e. children over early intervention age) found no significant effect. Results were more positive if the treatment group did not have receptive language difficulties, which are more likely to persist and more difficult to treat. Some positive results for receptive language skills were found if the interventions were targeted at specific areas, e.g. receptive vocabulary, word finding, comprehension of specific grammatical structures, etc. (Ebbels et al, 2016 pp.2-3). Ebbels et al found significant improvement on receptive and expressive language skills in primary and secondary aged students with DLD receiving 1:1 speech and language therapy (2016, pp.8-9).

Sansavini et al note the consensus in the literature on the importance of early intervention and diagnosis of DLD (Sansavini et al, 2021, p.14.). A 2021 systematic review of treatment studies found some evidence for the effectiveness of early intervention on some areas of language development. Early intervention effects last in the medium term for developing phonological skills but results of intervention targeting general language skills is mixed (Rinaldi, 2021, pp.18-19). For example, an Australian study by Wake et al compared the effect of home-based therapy sessions on children with language disorders with typically developing children. After 2 years they found language abilities for children in the treatment group normalised though they could not discern a significant effect of therapy sessions on most aspects of language development, including receptive and expressive language. Some effect was discerned for phonological awareness with a possible effect on reading ability (Wake et al, 2015, p.843).

## 5. Later diagnoses and autism

### 5.1 Age of first diagnosis

I was able to find only one study using Australian data that tracks age of first diagnosis for cohorts over 12 years old (Atherton et al, 2021) but this study was based on only 200 people. I am unable to give a good account of incidence of first diagnosis for the age groups requested.

Two systematic reviews have tracked age at first autism diagnosis between 1990 and 2019. Daniels and Mandell (2014) reviewed 42 papers published between 1990 and 2012. They provide a wide mean age range for first diagnosis at between 38 and 120 months. Van t' Hof et al (2021) analysed data from 56 studies and found a mean age for first diagnosis of 60.48 months (5 years) with a mean age range of between 30.9 months and 234.57 months (2021, p.862). The ranges provided by these reviews are significantly affected by age of participants in the studies reviewed. Many studies included only children, some studies included only older people. Daniels and Mandell use data from 12 countries. Van t' Hof et al use data from 40 countries. Both reviews include a single study from Australia (Daniels & Mandell, 2014, p.14-17; Van t' Hof, 2021, p.867).

International data indicates that age of diagnosis is decreasing (Daniels & Mandell, 2014, p.6; Sheldrick et al, 2017; Hanley et al, 2021). This contrasts with a recent UK-based study that found mean age of diagnosis rose from 9.6 years in 1998 to 14.5 years in 2018 (Russell et al, 2021, p.3). This might be explained by the fact that the Russell et al considered the entire UK population with an ASD diagnosis whereas the 2015 and 2021 systematic reviews included mostly studies of children. It may also be explained by regional differences in early intervention (Daniels & Mandell, 2014, p.10).

I have located 4 studies based on Australian data which discuss age of first diagnosis for ASD. Nassar et al was included in the Daniels and Mandell systematic review and focused on West Australian children between 2 and 8 years old. They found the mean age of first diagnosis decreasing from 4 years to 3 years throughout the 1990s (Nassar et al, 2009, p.1245). A study from Bent et al was included in the Van t' Hof systematic review and focused on children under 7 years. They found a mean age of first diagnosis of 49 months (Bent et al, 2015, p.318). May and Williams (2018) was not included in any of the reviews and looked at children under 13 years. Atherton et al was not included in any of the reviews and looked at 200 adults with ASD between 18 and 57 years.

According to May and Williams, the average age of diagnosis of children aged 0-12 years old is 6 years. The average is slightly higher in female children at 6.22 years. This estimate is based on Medicare data tracking first diagnosis item numbers from 2008 until 2016 and considers 73,463 children. The most frequent age of diagnosis is 5 until the year 2015/2016 when it lowers to 4 (May & Williams, 2018, p.5). In line with Russell et al (2021), May and Williams find that the rate of increase of older children being diagnosed is higher than the rate of increase for children under 5 (2018, pp.4-5). While this study underestimates total

prevalence due to limitations in the data, it likely captures most diagnoses occurring in this age range (p.2).

Based on a rough estimate obtained from a study by May and Williams (2018, p.4), for Australians diagnosed with ASD under the age of 12, 49% were diagnosed under the age of 6 and 51% were diagnosed between 6 and 12 years old. However, these shares will be significantly different when considering all those diagnosed with ASD in adolescence and adulthood. The average age of first diagnosis is bound to be higher than 6 when considering the entire population of Australians with ASD. This indicates that the average age of first diagnosis is above the early intervention age (>6) (Goodwin et al, 2018, p.2). This would be consistent with studies of other national populations. Atherton et al found the average age of diagnosis for their adult cohort was 15 for males and 21 for females (Atherton et al, 2021, p.4).

## 5.2 Reasons for later diagnosis

The rate of older people being diagnosed with autism is increasing. This appears to be true for adults (Russell et al, 2021, p.6) and older children (May & Williams, 2018, pp.4-5). Avlund et al. (2021) identify reasons that children may not receive an ASD diagnosis until later childhood or adolescence including:

- symptoms of other developmental disorder overshadow social impairments
- diagnostic threshold may not be met until it is clearer that the social demands on the child exceed their abilities
- the autistic symptoms may be expressed differently in early and later childhood
- socio-economic factors may influence the support a child receives (Avlund et al., 2021; Parikh et al, 2018).

A Melbourne based study also identifies limitations on resources as a primary reason that people do not receive a diagnosis until adolescence. They also note that symptoms being missed by the school system or primary care physician may result in missed diagnosis (Aggarwal & Angus, 2015, p.4).

International trends confirm that children are more likely to be diagnosed earlier if they have more severe autistic symptoms and more likely to be diagnosed later if they have milder autistic symptoms (Daniels & Mandell, 2014, p.7; Sheldrick et al, 2017 p.8; May & Williams, 2018, p.1; Parikh et al. 2018, p.6; Hanley et al, 2021, p.5; Avlund et al, 2021, pp.3849-3850). There is also some evidence to suggest that more severe symptoms can delay a diagnosis of autism if they are interpreted as symptoms of intellectual disability (Avlund et al, 2021, p.3851). A 2021 study by Atherton et al contrasts with the prevailing opinion, suggesting that people diagnosed later do not present differently but diagnoses may be missed due to environmental factors (Atherton et al, 2021, p.6). However, their results are also compatible with a worsening of symptoms over time in adults lacking proper diagnosis.

Barriers to adult autism diagnosis may include the following:

- there are few adult diagnostic screening tools
- difficulty remembering or recovering early developmental history
- limited understanding of adult autism in health professionals
- specialist multi-disciplinary team is often needed
- it requires significant time and effort from the patient
- symptoms of other conditions may mask autistic symptoms
- misdiagnosis or camouflaging of symptoms (Rødgaard et al, 2021, p.5; Scattoni et al, 2021, p.4130; Adamou et al, 2021, pp.1-2; Lai & Baron-Cohen, 2015; Legg et al, 2022, p.1).

There is mixed evidence to support these ideas. Rødgaard et al find that misdiagnosis or overshadowing of other childhood diagnoses may account for some of the reason autism diagnoses are missed. However, only 31% of males and 39% of females had childhood diagnoses at all, meaning that misdiagnosis or overshadowing cannot explain why diagnoses was not given in childhood for most later diagnosed people (2021, p.2).

A 2020 scoping review notes that factors prompting adult diagnosis include encouragement by parents or spouse, difficulties with social interaction or mental health issues (Huang et al, 2020).

### 5.3 Outcomes for people with later diagnoses

I could find only a single study that investigates quality of life for people diagnosed with autism later in life. Atherton et al found that people diagnosed earlier scored better on quality-of-life measures than people diagnosed later. Increasing age of diagnosis was correlated with increased social anxiety, social avoidance, and a lack of social support (2021, p.6).

Strong evidence suggests early intervention supports for children with ASD are effective in improving outcomes (Avlund et al, 2021, p.3843; Whitehouse et al, 2020; Productivity Commission, 2017; Clark et al, 2017, p.2; Zwaigenbaum et al, 2015, p.6; Estes et al. 2015). When children are diagnosed earlier, they have more access to services and interventions when their brains are most malleable. This means they can acquire skills from a younger age and build on these skills through their school years (Clark et al, 2017, pp.1-2).

Clark et al (2017) compared two groups of 7–9-year-olds with earlier or later diagnosis. The first group received diagnoses at 24 months. The second group received diagnoses between 3 and 5 years old. Those children diagnosed later received interventions later, received significantly less overall intervention, were slightly less likely to attend mainstream schooling, received more support at school age, had lower cognitive and language ability and were more likely to have an intellectual disability. These findings support the idea of improved outcomes for people diagnosed earlier and reduced functional capacity for people diagnosed later.

However, considering the ages of the comparison groups, this study may not reflect the outcomes for people diagnosed after early intervention age (<6 years). On the other hand, the underlying theory behind the effect is that earlier intervention works by making use of younger children's more malleable brains (Clark et al, 2017, pp.1-2; Anderson et al, 2014, p.8). If this is true then we may predict a similar trend for people diagnosed after early intervention age. As we found in [4.1 Later diagnosis of autism](#), this prediction is complicated by confounding factors such as multiple diagnoses for people with ASD, which may mean that they receive interventions targeting autistic symptoms even without a diagnosis of ASD.

Adults with autism typically have multiple diagnoses (Pelicano et al, 2020; Keller et al, 2020; Lai & Baron-Cohen, 2014). Adults with autism have an increased risk of depressive disorders, anxiety disorders, obsessive-compulsive disorder, attention deficit hyperactivity disorder, and personality disorders:

- more than 50% show increased depressive symptoms or depressive disorder
- as many as 66% report suicidal thoughts
- more than 50% may be diagnosed with anxiety disorders
- up to 40% may be diagnosed with attention deficit hyperactivity disorder
- up to 30% may be diagnosed with obsessive-compulsive disorder (Lai & Baron-Cohen, 2014, pp.1018-1019).

This information does not specify age of first diagnosis. However, there is evidence to suggest that later diagnosed people are more likely to have additional diagnoses (Daniels & Mandell, 2014; Goodwin et al, 2018; Pelicano et al, 2020; Rødgaard et al, 2021). A study of school age children by Goodwin et al notes that of people diagnosed between 5 and 18 years old, 58% had a psychiatric diagnosis. Of people diagnosed before 5 years old, only 29% had an additional diagnosis (Goodwin et al, 2018, p.4). In a small qualitative study of late diagnosed adults with autism, Pelicano et al note that of 28 participants in the study, 16 had at least one other psychiatric diagnosis and only 4 did not have any other medical condition (Pelicano et al, 2020, pp.21-23).

## 5.4 Supporting people with later life diagnoses

There is little research of the post-diagnostic needs of adults with ASD (Scatoni, 2021, p.2). Adults diagnosed with autism later in life have complex reactions and family, friends and clinicians supporting them should be aware of the potentially life-changing consequences of an adult diagnosis. In particular, later diagnosed adults and their caregivers report frustration with lack of post-diagnostic support (Legg et al, 2022, p.2; Scatoni et al, 2021, p.4142).

The UK's National Institute for Health and Care Excellence (NICE) has developed a series of clinical guidelines for people with autism. They recommend supports should be tailored to the person's age and developmental level ([NICE, 2021a](#), para. 1.3.1). However the

recommendations for support do not differ substantially for adults and young people except regarding their relative levels of autonomy and stages of life. For example, supported employment programmes ([NICE, 2021b](#), paras. 1.4.11-12) or residential care programmes (paras. 1.8.11-14) could be considered for adults with autism.

Considering the increased risk of co-morbid diagnoses as described in [5.1 Outcomes for people with autism](#), an increased focus on physical and mental health may be warranted. A 2020 systematic review by Benvenides et al found both cognitive behavioural therapy and mindfulness techniques had an emerging body of evidence as strategies for improving the health outcomes of older adults with autism. However, there is evidence that both strategies are also useful for children with autism (Benvenides et al, 2020, p.1351).

## 5.5 Effect of DSM-5 on ASD prevalence

For more information please refer to [RES 222 ASD diagnoses](#).

A 2019 systematic review investigated the effect of the changes to ASD diagnostic criteria between the DSM-IV-TR and the DSM-5. They found that approximately 1 in 5 people who would have received a diagnosis in DSM-IV-TR would not have received a diagnosis in the DSM-5. Further, only 28.8% of those who no longer meet ASD criteria would go on to meet diagnostic criteria for Social Communication Disorder (SCD) (Kulage et al, 2019, p.19). This means roughly 14% of people who met diagnostic criteria under DSM-IV no longer meet criteria for ASD or SCD. It is unclear what proportion of those people would go on to meet diagnostic criteria for other conditions and what proportion would remain below threshold for any DSM-5 diagnosis. According to this review, DSM-5 is contributing to a reduction in ASD diagnoses while the overall prevalence estimates continue to rise.

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## 7. Version control

Version	Amended by	Brief Description of Change	Status	Date
1.0	AHR908	Research paper on later life diagnoses for ASD and DLD	Final	15/02/2022

# Diagnoses of Autism Spectrum Disorder using the DSM-5

The content of this document is OFFICIAL.

**Please note:**

The research and literature reviews collated by our TAB Research Team are not to be shared external to the Branch. These are for internal TAB use only and are intended to assist our advisors with their reasonable and necessary decision-making.

Delegates have access to a wide variety of comprehensive guidance material. If Delegates require further information on access or planning matters they are to call the TAPS line for advice.

The Research Team are unable to ensure that the information listed below provides an accurate & up-to-date snapshot of these matters

**Research questions:**

1. What is the accuracy of Autism Spectrum Disorder diagnoses using the DSM 5, particularly for ASD levels 2 and 3 and particularly focussing on the interrater reliability of single discipline assessments?
2. What is the incidence of ASD diagnosis among family members? How likely is it that multiple siblings in a family will all have Autism Spectrum Disorder?
3. How has the rate of diagnosis of ASD changed since the publication of the DSM 5 diagnostic criteria?

**Date:** 10/12/2021

**Requestor:** Shannon Atkins

**Endorsed by (EL1 or above):** Shannon Atkins

**Cleared by:** Felicity Fallon

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## 2. Summary

This literature review addresses questions relating to the prevalence of Autism Spectrum Disorder (ASD). Findings include:

- ASD is strongly genetic. If someone has a family with ASD it is more likely that they will be diagnosed with ASD and it is more likely they will display autistic traits even if they don't meet the threshold for a diagnosis.
- DSM-5 diagnoses of ASD are overall more accurate than DSM-IV diagnoses. A true positive diagnosis is more likely if multiple assessment tools are used in the context of a multi-disciplinary team.
- The changes to DSM-5 ASD criteria likely reduced the frequency of ASD diagnoses, although prevalence continues to rise as a result of other factors.

These findings are provisional and may be altered with further research. Evidence supporting the high heritability of ASD is strong. Evidence is less reliable for prevalence estimates and accuracy of diagnoses. There is significant effort to understand the prevalence of ASD worldwide and to understand the effect of changes to the DSM-5 criteria. However, current studies are often marred by bias, lack of controls and small or unrepresentative samples. That being said, there is wide-spread consensus in the literature around the above findings.

## 3. Frequency of ASD diagnoses in families

Estimations of heritability of ASD range from 0.64 – 0.91, with some consensus emerging in the range 0.80 – 0.87 (Bai et al 2020; Sandin et al 2017; Tick et al 2016). High heritability means that for any two people, the more genes they share with each other, the more likely it is that they will share the highly heritable trait (Downes & Matthews, 2020). The closer the genetic relationship between a person with ASD and their relative, the more likely the relative will also have ASD. The literature notes recurrence rates of 80% for identical twins and 20% for non-identical siblings (Bai et al 2020; Girault et al 2020).

This is supported by population-based studies showing the likelihood of a person having ASD is increased if they have a family member with ASD (Girault et al 2020; Bai et al, 2020; Hansen et al 2019). One study predicts a 2-fold increase in likelihood of ASD diagnosis if you have a cousin with ASD and an 8-fold increase in likelihood of ASD diagnosis if you have an

older sibling with ASD (Hansen et al 2019). Girault et al (2020) also note that a sibling is even more likely to get a diagnosis of ASD if there are multiple people in the family with ASD.

Family members are also more likely to have more autistic traits (short of an ASD diagnosis) if someone in the family is diagnosed with ASD (Girault et al 2020; Page et al 2016). Girault et al also notes that a person with ASD getting a higher score on the Social Communication Questionnaire results in an increased chance of their sibling getting a diagnosis of ASD (Girault et al 2020).

## 4. Accuracy and inter-rater reliability of ASD diagnoses using DSM-5

While I was able to locate information establishing inter-rater reliability of DSM-5 ASD diagnoses, this should be treated with caution. The results do not come from studies that explicitly set out to study the accuracy of DSM-5 diagnoses. Studies examining other features of ASD or ASD diagnostic practices will often use inter-rater reliability to ensure study quality. In their study of ASD prevalence, Baio et al found 92.3% inter-rater agreement on presence or absence of ASD using DSM-5 criteria (2018, p.7). Taheri et al secured 100% inter-rater agreement for overall diagnosis and between 70% and 100% agreement on individual criteria (2014, p.118). In their study of gender differences in ASD diagnosis, Hiller, Young and Weber found substantial inter-rater agreement with Cohen's kappa scores of between 0.75 and 0.93 (2014, pp.4-5). Young and Rodi also secured strong inter-rater agreement for overall diagnosis with Cohen's kappa score of 0.91 (2014, p.761). These results demonstrate potential for high inter-rater agreement with DSM-5 ASD diagnoses, with somewhat lower agreement in individual criteria. They do not speak to accuracy of severity ratings (i.e. requiring support, requiring substantial support, requiring very substantial support).

Mazurek et al (2019) looked at use of severity ratings among clinicians. They found that assessment of severity levels of social communication and restrictive, repetitive behaviours using DSM-5 criteria largely agrees with other assessment tools as well as parental assessment of severity (p.7). However, they do point out a strong link between intelligence and severity ratings, which may mean that clinicians are conflating ASD symptoms with difficulties related to intellectual disability. Mazurek et al suggest that clinicians may be having difficulty:

“determining whether to assign ratings based on ASD symptom severity alone (more consistent with text examples) or based largely on need for support (more consistent with the level descriptors). If clinicians adhere to the latter interpretation, there may be greater potential for conflation of intellectual and symptom-related impairment. This poses problems for both inter-rater reliability and construct validity” (p.7).

Mazurek et al are also unaware of any studies looking at the inter-rater reliability of severity level assessments (p.8).

Hausman-Kedem et al looked at a group of 87 participants who had been diagnosed with ASD from psychologists or physicians in the community. They had predominately single-disciplinary diagnoses. Hausman-Kedem et al found the diagnoses did not hold up in 23% of cases when compared with best practice clinical estimates (2018, p.6). They also find that results of Autism Diagnostic Observation Schedule-2 (ADOS-2) substantially agrees with final best practice clinical estimates (2018, p.7). While the support for ADOS-2 is backed up by other studies, the discrepancy between community diagnoses and best practice clinical estimates is complicated by the participants' having DSM-IV diagnoses and the researchers using updated DSM-5 categories.

In their 2018 systematic review, Whigham et al found some support for diagnostic measures such as ADOS for adults, though they note that accuracy increases when multiple questionnaires and measures are used. They also observe that difficulties arise when distinguishing between ASD and some mental health conditions such as schizophrenia (p.15).

While there is better evidence to support tools used to diagnose ASD in children (Whigham, 2018, p.1), Randall et al found reason to be cautious about results supporting accuracy of diagnostic tools (2018, p.3). According to the evidence obtainable, ADOS scored highest for sensitivity and all tools assessed had similar results for specificity (p.2).

Further investigation will be required to provide a fuller picture of the overall accuracy of DSM-5 diagnoses and of tools based on DSM-5 diagnostic criteria. Despite some lack of confidence in the evidence, there is agreement in the literature that use of a variety of tools from a multi-disciplinary team gives the highest chance of correctly diagnosing a person with ASD.

## 5. Influence of DSM-5 ASD criteria on the prevalence of ASD

Autism prevalence rates are increasing (Taylor et al, 2020; Chiarotti & Venerossi, 2020; CDC 2020). The Autism and Developmental Disabilities Monitoring network (ADDM) estimates prevalence at 1 in 44 in sample United States communities (CDC 2020; Maener et al, 2021; Baio et al, 2018). Autism Spectrum Australia estimates prevalence at 1 in 70 in Australia (Autism Spectrum Australia, 2018). The reasons for the increase are likely to be complex and the exact proportion of the increase that is attributable to different factors is still a matter for debate. Kulage et al suggest:

“parental awareness and acceptance, less stigmatization, better trained clinicians, more thorough data collection methods, and even increasing genetic tendencies could be contributing factors. In addition, comorbid diagnoses are now allowable for ASD under DSM-5, enabling clinicians to give multiple comorbid diagnoses of intellectual disability, ASD, and ADHD, which could also explain why ASD rates have continued to rise since publication of the DSM-5” (Kulage et al, 2019, p.19).

Estimates of ASD prevalence are rising despite tightening diagnostic criteria in the current addition of the DSM-5. Since before publication of the DSM-5 there was concern about what the changes to ASD diagnostic criteria would do to ASD prevalence rates and especially whether people who failed to meet the new criteria would no longer be eligible for support (Kulage et al, 2019).

Kulage et al published a systematic review of the literature looking at the effect of the changes to ASD diagnostic criteria between the DSM-IV-TR and the DSM-5. They found that approximately 1 in 5 people who would have received a diagnosis in DSM-IV-TR would not have received a diagnosis in the DSM-V. Further, only 28.8 percent of those who no longer meet ASD criteria would go on to meet diagnostic criteria for Social Communication Disorder (SCD) (Kulage et al, 2019, p.19). This means roughly 14% of people who met diagnostic criteria under DSM-IV no longer meet criteria for ASD or SCD. It is unclear what proportion of those people would go on to meet other diagnostic criteria and what proportion would remain below threshold for any DSM-5 diagnosis.

According to this review, DSM-5 is contributing to a reduction in ASD diagnoses while the overall prevalence estimates continue to rise.

## 6. Literature Summary

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
<b>Inter-rater reliability of DSM-5 ASD Diagnoses</b>							
1	Mazurek et al 2019	Factors associated with DSM-5 severity level ratings for autism spectrum disorder	Autism, 23(2):468-476	To evaluate the use of these severity ratings for social communication and repetitive behaviour domains and to examine their relation to other measures of severity and clinical features.	Descriptive quantitative study of 248 children and adolescents with DSM-5 diagnoses. All participants received a non-standardized diagnostic clinical interview, standardized observation using the Autism Diagnostic Observation Schedule–Second Edition (ADOS-2), cognitive assessment, and assessment of behavioral functioning. Participants were assessed by a psychologist, physician or multi-disciplinary team.	Higher severity ratings in both domains were associated with younger age, lower intelligence quotient, and greater Autism Diagnostic Observation Schedule–Second Edition domain-specific symptom severity. Greater restricted and repetitive behavior severity was associated with higher parent-reported stereotyped behaviours. Severity ratings were not associated with emotional or behavioural problems. Strong associations between	The study is based on a large sample that appears representative in terms of gender and functioning. No significant bias was detected however the clinicians undertaking the assessments are specialists in ASD diagnosis and may not be representative of clinicians in the community.

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
						intelligence quotient and DSM-5 severity ratings in both domains suggest that clinicians may be including cognitive functioning in their overall determination of severity.	
2	Hausman-Kedem et al 2018	Accuracy of Reported Community Diagnosis of Autism Spectrum Disorder	Journal of Psychopathology and Behaviour Assessment. 40(3): 367–375.	To compare community diagnoses of Autism Spectrum Disorder (ASD) reported by parents to consensus diagnoses made using standardized tools plus clinical observation.	87 participants (85% male, average age 7.4 years), with reported community diagnosis of ASD were evaluated using the Autism Diagnostic Observation Schedule) (ADOS-2), Differential Ability Scale (DAS-II), and Vineland Adaptive Behaviour Scales (VABS-II). Detailed developmental and medical history was obtained from all participants. Diagnosis was based on clinical consensus of at	23% of participants with a reported community diagnosis of ASD were classified as non-spectrum based on our consensus diagnosis. Participants enrolled with community diagnosis of PDD-NOS were significantly more likely to be classified as non-spectrum on the study consensus diagnosis than Participants with Autism or Asperger. This study shows	The sample size is small and males are over-represented. Consensus diagnoses made using DSM-5 criteria were compared to community diagnoses using DSM-IV criteria. Results may reflect changes in criteria as well as differences between community diagnosis and consensus diagnosis.

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
					least two expert clinicians, using test results, clinical observations, and parent report.	suboptimal agreement between community diagnoses of ASD and consensus diagnosis using standardized instruments.	
3	Wigham et al 2019	Psychometric properties of questionnaires and diagnostic measures for autism spectrum disorders in adults: A systematic review	Autism, 23(2): 287-305	Systematic review of research evidence on structured questionnaires and diagnostic measures for adults with Autism published since 2014.	Systematic review	Limited evidence for accuracy of structured questionnaires. Sensitivity and specificity of structured questionnaires were best for individuals with previously confirmed ASD and reduced in participants referred for diagnostic assessments, with discrimination of ASD from mental health conditions especially limited. For adults with intellectual disability, diagnostic accuracy increased when a combination of structured questionnaires were	Design of included studies were case-control, cross sectional or retrospective, making comparison of results difficult. Case-control studies are at risk of bias which limits to relevance of the reviews results. However, the authors point out that both stronger and weaker studies agreed on the poor psychometric properties of the tools investigated.

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
						used. In mental health settings, the use of a single structured questionnaire is unlikely to accurately identify adults without autism spectrum disorder or differentiate autism spectrum disorder from mental health conditions.	
4	Randall et al	Diagnostic tests for autism spectrum disorder (ASD) in preschool children	Cochrane Database of Systematic Reviews	To identify which diagnostic tools, including updated versions, most accurately diagnose ASD in preschool children when compared with multi-disciplinary team clinical judgement. To identify how the best of the interview tools compare with CARS, then how CARS compares with ADOS: which ASD diagnostic tool - among ADOS, ADI-R, CARS, DISCO, GARS, and 3di - has the best diagnostic test	Systematic review	ADOS scored a summary sensitivity of 0.94 and a summary specificity of 0.80. When compared with other assessed tools, ADOS scored highest for sensitivity and all tools had similar results for specificity.	Studies reviewed showed some risk of bias though studies at high risk of bias were excluded. Overall, authors advice to interpret results with caution due to sample sizes of included studies and potential conflicts of interest.

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
				accuracy?; is the diagnostic test accuracy of any one test sufficient for that test to be suitable as a sole assessment tool for preschool children?; is there any combination of tests that, if offered in sequence, would provide suitable diagnostic test accuracy and enhance test efficiency?; if data are available, does the combination of an interview tool with a structured observation test have better diagnostic test accuracy (i.e. fewer false-positives and fewer false-negatives) than either test alone?			
<b>Frequency of ASD diagnoses in families</b>							
1	Bai et al Sept 2020	Inherited Risk for Autism Through Maternal and Paternal Lineage	Biological Psychiatry; 88:480–487	Review data on frequency of ASD among family members	Quantitative correlational study using data from the Swedish National Patient Register and the Multi-Generation Register	1.55% of children in the cohort were diagnosed with ASD. Among their maternal /paternal aunts and uncles 0.24% and 0.18%	The sample is large (847,732 children in total and 13,103 diagnosed with ASD) and so results are robust. However the

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
					for a cohort of children born between 2003 and 2012. Researchers compared frequency of ASD diagnosis with family relations and sex in a group of 847,732 children.	were diagnosed with ASD, respectively. Offspring of mothers with a sibling(s) diagnosed with ASD had higher rates of ASD than the general population (relative risk, 3.05; 95% confidence interval, 2.52–3.64). These findings establish a robust general estimate of ASD transmission risk for siblings of individuals affected by ASD, the first ever reported. Our findings do not suggest female protective factors as the principal mechanism underlying the male sex bias in ASD.	sample is drawn entirely from Swedish national registers and so may not be wholly applicable to other national contexts (depending on variation in diagnostic habits). Also, diagnoses are made using ICD 8,9, and 10. Results may be different using DSM-5 diagnoses.
2	Girault et al 2020	Quantitative trait variation in ASD probands and toddler sibling outcomes at 24 months	Journal of Neurodevelopmental Disorder 12:5	To investigate how quantitative variation in ASD traits and broader developmental domains in older siblings with ASD	Compared 385 pairs of toddlers and their older siblings using data from the Infant Brain Imaging	Older siblings' scores on the Social Communication Questionnaire predicts whether	The study uses a substantial sample of 385 sibling pairs. However, the study uses DSM-IV to diagnose toddlers

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
				(probands) may inform outcomes in their younger siblings.	Study. Toddlers and older siblings were each assessed using age appropriate diagnostic and adaptive behaviour assessment tools to determine presence of ASD and autistic traits.	younger siblings will receive an ASD diagnosis. There is large variation in autistic traits exhibited by siblings. However, the severity of autistic traits in the older sibling predicts severity in the younger sibling.	as the IBIS data was gathered before the DSM-5 a was released. The study did not consider if this would have an impact on results. Also, while age-appropriate clinical tools were used in the assessment of the subjects, the different tools casts some doubts on the comparison between older and younger siblings.
3	Hansen et al 2019	Recurrence risk of autism in siblings and cousins: a multi-national population-based study	Journal of the American Academy of Child and Adolescent Psychiatry, 58(9): 866–875	To estimate ASD recurrence risk among siblings and cousins by varying degree of relatedness and by sex	International population-based cohort study of children born 1998–2007. Follow up 2011–2015. Subjects were monitored for an ASD diagnosis in their older siblings or cousins (exposure) and for their own ASD diagnosis (outcome). The relative recurrence	Research found an 8.4-fold increase in the risk of ASD following an older sibling with ASD and a 17.4-fold increase in the risk of Childhood Autism (CA) following an older sibling with CA. A 2-fold increase in the risk for cousin recurrence was observed for both disorders.	Very large sample of almost 9 million children (with 29,998 cases of ASD and 33,769 cases of childhood autism). Measures both shared genetic and non-genetic factors. There was missing parental information in only a small proportion of the sample. Results are robust.

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
					risk was estimated for different sibling- and cousin-pairs, for each site separately and combined, and by sex	Researchers also found a significant difference in sibling ASD recurrence risk by sex.	
4	Sandin et al 2017	The Heritability of Autism Spectrum Disorder	Journal of the American Medical Association; 318(12): 1182-1184	To calculate the heritability of ASD by reanalysing a previous data set.	Sample of 3,557,446 pairs of siblings was examined for presence of ASD. Total of 14,516 children were diagnosed with ASD. Liability threshold models were used to identify additive and non-additive genetic factors, shared and non-shared environmental factors.	On one model comparing on heritability and non-shared environmental factors, heritability was estimated at 0.87. On a model with all 4 factors, heritability was 0.69. Using only twins in the sample, heritability was 0.87. The heritability of ASD is high and the risk of ASD increased with increasing genetic relatedness.	The sample is very large but taken from only Swedish sample and so may not be wholly applicable to other national contexts. Frequency of ASD diagnoses in the sample (<0.5%) is far below incidence in the general population (1-2%). This study focusses on heritability and may not reflect other familial factors.
5	Page et al 2016	Quantitative autistic trait measurements index background genetic risk for ASD in Hispanic families	Molecular Autism 7:39	To fill a gap in the literature by investigating the relationship of quantitative autistic traits (QAT) to liability of ASD in an example	Researchers examined QAT scores in siblings and parents of 83 Hispanic children with ASD, and 64 non-ASD controls, using the Social	Measured correlations (between children with ASD and i) first degree relative, ii) unaffected first degree relatives in ASD affected	Small sample relative to these types of studies and while the study depends on a less heterogeneous sample than other studies (Hispanics),

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
				non-Caucasian population.	Responsiveness Scale-2.	families and iii) spouses) supported previous studies of non-Hispanic populations.	it also i) restricts its conclusions to this population group; and ii) shows how the results for this group coincide with studies of other population groups.
6	Tick et al 2016	Heritability of autism spectrum disorders: a meta-analysis of twin studies	Journal of Child Psychology and Psychiatry 57:5; 585-595	To assess the evidence of environment and genetic factors in the aetiology of ASD	Systematic review and meta-analysis of all ASD twin studies.	ASD heritability estimates were 64–91%. Shared environmental effects became significant as the prevalence rate decreased from 5–1%: 07–35%.	Results are robust. The review contains a meta-analysis of twin studies, which are the standard for heritability studies. Authors have also explained discrepancy between the results of the meta-analysis and previous studies, namely, an over-estimation of the significance of environmental factors was due to some previous studies' overinclusion of non-identical twins in the samples.
7	Frazier et al 2015	Quantitative autism symptom patterns recapitulate differential	Molecular Autism	To establish the extent to which family transmission pattern	Researchers analysed data from 5515 siblings (2657 non-ASD and 2858	Non-ASD children manifested elevated ASD symptom burden when they	

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
		mechanisms of genetic transmission in single and multiple incidence families		and sex modulate ASD trait aggregation	ASD). Autism symptom levels were measured using the Social Responsiveness Scale (SRS) and by computing DSM-5 symptom scores based on items from the SRS and Social Communication Questionnaire.	were members of multiple incidence families—this effect was accentuated for male children in female ASD-containing families—or when they had a history of language delay with autistic qualities of speech. Recurrence risk for ASD was higher for children from female ASD-containing families than for children from male-only families	
<b>Influence of DSM-5 ASD criteria on the prevalence of ASD</b>							
1	Kulage et al 2019	How has DSM-5 Affected Autism Diagnosis? A 5-Year Follow-Up Systematic Literature Review and Meta-analysis	Journal of Autism and Developmental Disorders	To 1) determine the change in frequency of ASD diagnosis in the first five years after publication of the revised DSM-5 ASD criteria; (2) identify the DSM-IV-TR autism subtypes most affected by the new criteria; and (3) assess the potential of an alternative	Systematic review using PRISMA guidelines. Qualitative and quantitative meta-analysis of 33 published articles.	Using a random effects model, the pooled proportion suggests a 20.8% reduction in ASD diagnoses. Pooled effects suggest statistically significant reductions in ASD diagnoses of 10.1% for those with AD	The study is of high quality as a systematic review and meta-analysis, although the underlying data has a moderate risk of bias stemming from lack of masking of raters to results of the references standard, DSM-IV-

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
				diagnosis of SCD for individuals who meet DSM-IV-TR but not DSM-5 ASD diagnostic criteria		and 23.3% for those with Asperger's Disorder when DSM-5 criteria were applied. The reduction in diagnoses for PDD-NOS was not statistically significant. Less than one-third [28.8%] of those who met DSM-IV-TR ASD diagnostic criteria but not DSM-5 would meet SCD diagnostic criteria.	TR diagnosis, and failure to assess interrater agreement in classification of DSM-5 diagnoses. Findings should be interpreted with caution however this study does represent the most comprehensive exploration of the data available.
2	Baio et al 2018	Prevalence of Autism Spectrum Disorder Among Children Aged 8 Years — Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2014	Centre for Disease Control and Prevention – Morbidity and Mortality Weekly Report – Surveillance Summaries 67(6)	To determine ASD prevalence in 11 communities in the United States.	The first phase involves review and abstraction of comprehensive evaluations that were completed by professional service providers in the community. In the second phase of the study, all abstracted information is reviewed systematically by experienced	For 2014, the overall prevalence of ASD among the 11 ADDM sites was 16.8 per 1,000 (one in 59) children aged 8 years.	Sample size is adequate to draw conclusion about estimated prevalence in the age and communities studied. However, ADDM study is sometimes used as an estimate of prevalence for the entire United States. Samples chosen are not representative of

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
					clinicians to determine ASD case status.		the entire US. Did not specify whether raters were aware of the findings of other raters.
3	Taheri, Perry, Factor 2014	A Further Examination of the DSM-5 Autism Spectrum Disorder Criteria in Practice	Journal on Developmental Disability 20(1)	To determine whether children and adolescents diagnosed with Autistic Disorder or PDD-NOS on DSM-IV criteria would continue to meet DSM-5 ASD criteria. To replicate and extend the findings of an earlier paper in a different sample of older individuals with lower cognitive and adaptive skills	File review of 22 children and adolescents previously diagnosed under DSM-IV criteria. Records were then reassessed using DSM-5 criteria.	Only 55% of the sample met the DSM-5 criteria for ASD; this included 69% of those who had an original DSM-IV-TR diagnosis of AD, and only 17% (one child) with an original diagnosis of PDD-NOS.	Reassessments were completed using DSM-5 checklist rather than clinical diagnoses. Children diagnosed with Aspergers were excluded. Small sample size although study intentionally worked as an extension of a previous study with an adequate sample size. Although masking of participants occurred, the study did not specify whether raters were aware of the findings of other raters.
4	Hiller, Young and Weber 2014	Sex Differences in Autism Spectrum Disorder based on DSM-5 Criteria: Evidence from	Journal of Abnormal Child Psychology	To explore sex differences in the behavioural presentation of girls and boys diagnosed	Quantitative descriptive study of 138 children with ASD. Diagnoses were provided by two clinicians and	While no sex differences were found in the broad social criteria presented in the DSM-IV-TR or	Adequate sample size and reported inter-rater reliability between clinicians but study did not specify whether

	Author / Date	Title	Source	Aim / Objective	Methods	Results	Quality of Research
		Clinician and Teacher Reporting		with high-functioning ASD.	then statistical analyses were applied.	DSM-5, numerous differences were evident in how boys and girls came to meet each criterion.	raters were aware of the findings of other raters.
5	Young and Rodi 2014	Redefining Autism Spectrum Disorder Using DSM-5: The Implications of the Proposed DSM-5 Criteria for Autism Spectrum Disorders	Journal of Autism and Developmental Disorders 44:758–765	To compare overlap of DSM-IV pervasive development delay diagnoses and DSM-5 autism diagnoses.	223 subjects who were either referred for a DSM-IV diagnosis and did not receive one, or who received a DSM-IV diagnoses were reassessed using DSM-5 criteria.	Of the 210 participants in the present study who met DSM-IV TR criteria for a PDD only 57.1 % met DSM-5 criteria for autism spectrum disorder when criteria were applied concurrently during diagnostic assessment	Adequate sample size and reported inter-rater reliability between clinicians but study did not specify whether raters were aware of the findings of other raters. DSM-5 diagnoses were completed by one or two clinicians and so did not meet best practice guidelines for clinical assessments.

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## 8. Version control

Version	Amended by	Brief Description of Change	Status	Date
0.1	AHR908	Literature review on the incidence and reliability of ASD diagnoses using DSM-5 criteria.	Draft	10-12-21
1.0	FFM634	Final	Completed	10-12-21