

October 2025



Avoiding Diagnostic Overshadowing when Caring for Adults with Down Syndrome

Author

Brian Chicoine, MD

Reviewers

Priya Chandan, MD, PhD, MPH

George Capone, MD

Introduction

Individuals with Down syndrome and their families report that they are often told a presenting symptom is, “just Down syndrome” rather than a co-occurring condition. The Joint Commission describes this as *diagnostic overshadowing*, defined as “the attribution of symptoms to an existing diagnosis rather than a potential co-morbid condition.”¹ The term first appeared in the English-language research literature in the early 1980s to describe the misattribution of symptoms to an underlying intellectual disability rather than to a psychologic or psychiatric diagnosis. Despite increasing recognition of diagnostic overshadowing, there remains a tendency to assess individuals with an intellectual disability less accurately,² leading to underdiagnosis or missed diagnoses of co-occurring health conditions such as pain, depression,

psychosis, adjustment disorder, hypothyroidism, or severe sleep apnea. While diagnostic overshadowing was initially applied to those with an underlying intellectual disability, it is recognized today as a problem in the treatment of people with other underlying conditions such as mental illness,^{2,3} neurodevelopmental disabilities,⁴ and movement disorders.⁵

In diagnostic overshadowing as it pertains to Down syndrome, when the healthcare professional focuses primarily or only on the Down syndrome diagnosis and attributes many or all symptoms to Down syndrome alone, co-occurring conditions can be overlooked. The related term “treatment overshadowing” is also used.³ This refers to the treatment phase, rather than the diagnostic phase. In either case, an opportunity is missed to provide effective interventions that would improve quality of life.

Diagnostic Overshadowing

Diagnostic overshadowing can be attributed to a variety of biases:

Anchoring bias: the tendency to focus or rely only or most stringently on a particular piece of data or information

Premature closure: discontinuing the evaluation or data-collection process before all the information is known

Implicit bias: personal feelings, emotions, or beliefs that can sway assessment and decision-making

Cognitive Biases to Avoid

Diagnostic overshadowing can be attributed primarily to lack of familiarity with individuals with Down syndrome and the medical comorbidities to which they are prone. Practitioners should also be aware of various cognitive biases.^{2,6}

- **Anchoring bias:** the tendency to focus or rely primarily on a particular piece of data or information
- **Premature closure:** discontinuing the evaluation or data-collection process before all the information is known
- **Implicit biases:** feelings, emotions, or beliefs that can sway one's assessment and decision-making

One additional finding regarding diagnostic overshadowing is of note. In one study, it was found that the physicians most vulnerable to diagnostic overshadowing were those with the lowest and those with the highest level of experience.⁶ Thus, awareness of diagnostic overshadowing is important even for seasoned clinicians.

An important factor for individuals with Down syndrome is practitioners' reluctance to make patient-specific accommodations for appropriate care. For example, the "burden of care" for an individual with Down syndrome might include involving an anesthesiologist in a procedure, which might not be considered for

an individual without Down syndrome. When a practitioner decides to exclude the individual with Down syndrome from that testing or treatment because of the perceived burdens or risks, without having a shared decision-making discussion, this is overshadowing. To minimize this bias, practitioners should engage in shared decision-making discussions regarding the recommended diagnostic and treatment approaches. Short-term difficulties should not bar individuals with Down syndrome from the long-term benefits of appropriate care.

Diagnostic Overshadowing Due to Co-Occurring Conditions

In addition to diagnostic overshadowing due to the diagnosis of Down syndrome, individuals with Down syndrome also experience diagnostic overshadowing for other reasons.

Mental Health and/or Behavioral Conditions

As noted in the [*Common Characteristics of Adults with Down Syndrome*](#) article, it is important to understand the interaction between mental and physical health in individuals with Down syndrome.^{7,8} When unable to verbally describe their physical symptoms, they may "communicate" those symptoms via observable behaviors. Thus, any sudden and/or significant change in a person's baseline behavior, communication, or adaptive function requires further medical investigation by a knowledgeable clinician.

Alzheimer's Disease

Individuals with Down syndrome are at greater risk for Alzheimer's disease, and at a younger age (see the article [Alzheimer's Disease in Adults with Down Syndrome](#)). Although symptoms of Alzheimer's disease are rare before age 40, it is often assumed that an individual with Down syndrome who develops a cognitive-adaptive skill loss has Alzheimer's disease, even at a young age.⁹ In addition to the greater risk of Alzheimer's disease, other factors contribute to the assumption that Alzheimer's disease is the cause for any type of cognitive decline in adults with Down syndrome. The symptoms of Alzheimer's disease may be similar to those of other conditions causing a cognitive decline, such as what is now often referred to as Down Syndrome Regression Disorder,¹⁰ which tends to affect adults between 15-25 years of age. In addition, there are clinical limitations and other difficulties diagnosing Alzheimer's disease in individuals with Down syndrome.¹¹

Physiological Features Unique to Down Syndrome

Individuals with Down syndrome may have reduced function in multiple organ systems.¹¹ For example, their immune systems may be affected by lower white blood cell counts, differences in B-cell and T-cell function, and other factors.^{12,13} Likewise, they may have reduced kidney function due to a smaller number of nephrons, exacerbated by poor hydration.¹⁴ Accepting the inevitability of the dysfunction without

addressing possible remedies is another form of diagnostic overshadowing.¹⁵⁻¹⁷ Using kidney function as an example, limiting medications that can further impair kidney function and promoting good hydration are interventions that can reduce the effect of this common feature.

Common Behavioral Characteristics

In the article [Common Characteristics of Adults with Down Syndrome](#), behaviors typical in individuals with Down syndrome were described. Although departures from these behaviors may signify an underlying mental health problem, it is often a response to negative or stressful life events. When such behaviors are assumed to represent a mental illness rather than a common characteristic,¹⁸ this can lead to misdirected evaluation, diagnostic, and treatment decisions.

Autism

It is now recognized that an individual with Down syndrome can also have autism,^{19,20} and that autism is more common in individuals with Down syndrome. Autism and Down syndrome have overlapping features affecting behavior, communication, and social skills, making it difficult to diagnose autism in young children with Down syndrome.¹⁹ However, failure to consider the diagnosis of autism can delay or prevent beneficial therapies.

Post-Mortem Diagnostic Overshadowing

Diagnostic overshadowing post-mortem has been studied and has been reported as common in individuals with Down syndrome and other intellectual disabilities.²¹ In this type of overshadowing, the diagnosis of the disability is listed as the cause of death, whereas other contributing factors or diagnoses are either not assessed or not provided on the death certificate. When an easily-recognizable condition such as Down syndrome is present, the cause of death may be attributed to this only, and a less thorough assessment provided than for someone without Down syndrome.²² If the cause of death is uncertain, this further increases the likelihood that the death will be attributed to the disability.²³ This incomplete assessment of the cause of death biases our understanding of co-occurring conditions associated with morbidity and mortality in this population.

Summary

Diagnostic overshadowing in individuals with Down syndrome involves misattributing important symptoms to the syndrome itself, without considering possible co-occurring conditions. It also may involve over-attributing certain behavioral and cognitive traits to conditions for which this population is at greater risk, such as Alzheimer's disease. In whatever form, diagnostic overshadowing may result in missed opportunities for diagnosis and treatment in individuals with Down syndrome.

Resources

- 1 Joint Commission. Sentinel Event Alert 65: Diagnostic overshadowing among groups experiencing health disparities. <https://www.jointcommission.org/resources/sentinel-event/sentinel-event-alert-newsletters/sentinel-event-alert-65-diagnostic-overshadowing-among-groups-experiencing-health-disparities/>. Accessed June 22, 2024.
- 2 Hallyburton A. Diagnostic overshadowing: An evolutionary concept analysis on the misattribution of physical symptoms to pre-existing psychological illnesses. *Int J Ment Health Nurs*. 2002;31(6): 1360–1372. doi:10.1111/inm.13034
- 3 Jones S, Howard L, Thornicroft G. 'Diagnostic overshadowing': worse physical health care for people with mental illness. *Acta Psychiatr Scand*. 2008;118(3):169-171. doi:10.1111/j.1600-0447.2008.01211.x
- 4 Iezzoni LI. Dangers of diagnostic overshadowing. *N Engl J Med*. 2019;380(22):2092-2093. doi:10.1056/NEJMp1903078
- 5 Agaronnik ND, El-Jawahri A, Iezzoni LI. Perspectives of patients with pre-existing mobility disability on the process of diagnosing their cancer. *J Gen Intern Med*. 2021;36(5):1250-1257. doi:10.1007/s11606-020-06327-7
- 6 Almeida B, Samouco A, Grilo F, Pimenta S, Moreira AM. Prescribing stigma in mental disorders: a comparative study of Portuguese psychiatrists and general practitioners. *Int J Soc Psychiatry*. 2022;68(4):708-717. doi:10.1177/00207640211002558
- 7 Lazris A, Roth A, Haskell H, James J. Diagnostic overshadowing: when cognitive biases can harm patients. *Am Fam Physician*. 2023;108(3):292-294
- 8 Molloy R, Munro I, Pope N. Understanding the experience of diagnostic overshadowing associated with severe mental illness from the consumer and health professional perspective: a qualitative systematic review protocol. *JBIM Evid Synth*. 2021;19(6):1362-1368. doi:10.11124/JBIES-20-00244
- 9 Tsou AY, Bulova P, Capone G, et al. Medical care of adults with Down syndrome: a clinical guideline. *JAMA*. 2020;324(15):1543-1556. doi:10.1001/jama.2020.17024
- 10 Worley G, Crissman BG, Cadogan E, Milleson C, Adkins DW, Kishnani PS. Down syndrome disintegrative disorder: new-onset autistic regression, dementia, and insomnia in older

- children and adolescents with Down syndrome. *J Child Neurol.* 2015;30(9):1147-1152. doi:10.1177/0883073814554654
- 11 Chicoine B, Rivelli A, Fitzpatrick V, Chicoine L, Jia G, Rzhetsky A. Prevalence of common disease conditions in a large cohort of individuals with Down syndrome in the United States. *J Patient Cent Res Rev.* 2021;8(2):86-97. doi:10.17294/2330-0698.1824
 - 12 Dieudonné Y, Uring-Lambert B, Jeljel MM, et al. Immune defect in adults with Down syndrome: insights into a complex issue. *Front Immunol.* 2020;11:840. doi:10.3389/fimmu.2020.00840
 - 13 Bordon Y. Immune dysregulation in Down syndrome. *Nat Rev Immunol* 2023; 23:201. doi:10.1038/s41577-023-00855-z
 - 14 Mercer ES, Broecker B, Smith EA, Kirsch AJ, Scherz HC, Massad CA. Urological manifestations of Down syndrome. *J Urol.* 2004 ;171(3):1250–3. doi: 10.1097/01.ju.0000112915.69436.91
 - 15 Emes D, Hüls A, Baumer N, et al. COVID-19 in children with Down syndrome: data from the Trisomy 21 Research Society survey. *J Clin Med.* 2021;10(21):5125. doi:10.3390/jcm10215125
 - 16 Ghezzi M, Garancini N, De Santis R, et al. Recurrent respiratory infections in children with Down syndrome: a review. *Children (Basel).* 2024;11(2):246. doi:10.3390/children11020246
 - 17 Kusters MA, Manders NC, de Jong BA, van Hout, RW, Rijkers GT, de Vries E. Functionality of the pneumococcal antibody response in Down syndrome subjects. *Vaccine.* 2013;31(52):6261 – 6265. doi:10.1016/j.vaccine.2013.09.070
 - 18 Hurley AD. The misdiagnosis of hallucinations and delusions in persons with mental retardation: a neurodevelopmental perspective. *Semin Clin Neuropsychiatry.* 1996;1(2):122-133. doi:10.1053/SCNP00100122
 - 19 Diniz NLF, Parlato-Oliveira E, Pimenta PGA, Araújo LA, Valadares ER. Autism and Down syndrome: early identification and diagnosis. *Arq Neuropsiquiatr.* 2022;80(6):620-630. doi:10.1590/0004-282X-ANP-2021-0156
 - 20 Spinazzi NA, Santoro JD, Pawlowski K, et al. Co-occurring conditions in children with Down syndrome and autism: a retrospective study. *J Neurodev Disord.* 2023;15(1):9. doi:10.1186/s11689-023-09478-w
 - 21 Landes SD, Stevens JD, Turk MA. Postmortem diagnostic overshadowing: reporting cerebral palsy on death certificates. *J Health Soc Behav.* 2022;63(4):525-542.

doi:10.1177/00221465221078313

- 22 Baird PA, Sadovnick AD. Underlying causes of death in Down syndrome: accuracy of British Columbia death certificate data. *Can J Public Health*. 1990;81(6):456-461.
- 23 Landes SD, Turk MA, Finan JM. Factors associated with the reporting of Down syndrome as the underlying cause of death on US death certificates. *J Intellect Disabil Res*. 2022;66(5):454-470. doi:10.1111/jir.12926