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# Diagnostic Overshadowing: A Delayed Diagnosis of Autism Spectrum Disorder in a Child who is Deaf

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**Abstract**

Diagnostic overshadowing occurs when a patient with a pre-existing diagnosis presents with symptoms that are attributed to the diagnosis, but actually reflect a separate issue. This diagnostic overshadowing can lead to delays in diagnosis, as well as disparities in health care and outcomes. The following case will provide an example in which diagnostic overshadowing contributed to the delay of an autism diagnosis in a child who is deaf. With 40 to 50% of children who are deaf or hard of hearing having at least one co-existing medical/developmental condition, and the current prevalence of autism at 1 in 36 children in the United States, the intersection of these conditions is substantial. Pediatricians, otolaryngologists, audiologists, and speech therapists are all likely to encounter young children who are deaf or hard of hearing and in the process of acquiring language. Using interdisciplinary collaboration can support identification of children with coexisting conditions. Clinicians can benefit from increased familiarity with the presentation of children who are both deaf and neurotypical, and how it tends to differ from children with autism, reducing the chance of delayed or incorrect diagnosis and strengthening the formation of care plans.

**Keywords:** autism, deaf, diagnostic overshadowing, hearing loss, language development

**Acronyms:** ASD = Autism Spectrum Disorder; EHDI = early hearing detection and intervention; DHH = deaf or hard of hearing

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Diagnostic overshadowing occurs when a patient with a pre-existing diagnosis presents with symptoms that are attributed to the diagnosis, but truly reflect a separate issue. When considering a diagnosis of autism in an individual who also has a sensory issue (deaf or hard of hearing, low vision, etc.), diagnostic overshadowing can occur in two distinct manners: *misdiagnosis* of autism (symptoms that are attributed to autism but are actually due to the sensory diagnosis), and *missed diagnosis* of autism (symptoms that are attributed to the sensory diagnosis when they are actually due to autism; Ludwig et al., 2022). Discerning whether a child is deaf or hard of hearing and/or has autism provides some diagnostic challenges. Most states in the United States currently have a formal early hearing detection and intervention (EHDI) program that oversees newborn hearing screening and follow-up policies (National Center for Hearing Assessment and Management, 2024). EHDI programs have contributed to children who are deaf or hard of hearing being identified earlier in life, and often as infants if they are congenitally deaf or hard of hearing. A reliable diagnosis of autism can be made by an experienced professional starting around 2 years of age, although children with a milder presentation may not raise clinical suspicion until they are older (Centers for Disease Control and Prevention, 2022). Thus, for the majority of children who are congenitally deaf or hard of hearing, identification of hearing status will be well established prior to the time suspicion for possible autism may arise. This has the potential to confound the identification of autism due to some shared presenting (but etiologically differing) symptoms, such as delayed or lack of language development, reduced responsiveness, and diminished social engagement, all of which may be inappropriately attributed to lack of hearing and access to spoken language. The following case will provide an example of diagnostic overshadowing that contributed to the delay of an autism diagnosis in a child who is deaf.

## Case Report

### History

A 2-year, 8-month-old female who had been diagnosed as deaf presented to our specialty clinic for deaf and hard of hearing children. She had been identified with late onset profound bilateral sensorineural hearing loss by an outside

pediatric otolaryngologist at 14 months of age after concerns for lack of receptive and expressive language development in both spoken English and sign language. Review of available records from her otolaryngologist (who was part of a pediatric cochlear implant program) included her diagnosis of bilateral profound sensorineural hearing loss, a Magnetic Resonance Imaging (MRI) scan of her internal auditory canals which was normal and specifically made reference to normal cochlear development and normal caliber cochlear nerves, a statement of her having been determined to be a good candidate for cochlear implant, and her intra-operative record. Copies of her aided and unaided audiogram assessment prior to cochlear implantation were unfortunately not provided for direct review. Parents reported that she had not benefited substantially from traditional hearing aid amplification. At 22 months old, she had bilateral cochlear implants placed at the outside pediatric cochlear implant program that housed the pediatric otolaryngologist and audiology team that had been seeing her.

She was enrolled in regular occupational therapy for sensory defensiveness, as well as twice weekly auditory verbal speech therapy, which had been initiated prior to cochlear implantation. An initial speech therapy evaluation done when she was two years old included the Receptive Expressive Emergent Language Scale, Third edition (REEL-3) assessment, which demonstrated receptive language, expressive language, and language ability scores in the *extremely low* range (standard scores [SS] all < 55). Follow-up re-evaluation included re-administration of the REEL-3 ten months later, and standard scores had changed marginally (receptive language SS = 55, expressive language SS = 57, language ability SS = 55). Diagnosis at both assessments was noted to be consistent with a mixed expressive/receptive language disorder. Additionally, her family had routine exposure to sign language learning through a deaf education parent advisor via Early Childhood Intervention who worked with them weekly. Observation of what the family was using with her during our visit was most consistent with a CASE model (Conceptually Accurate Signed English) using signs based on conceptual meaning consistent with American Sign Language but using an English-based syntax and grammar system.

Parents reported that for several months, she had been increasingly refusing to wear her cochlear implant processors, and they were concerned with lack of progress with language development (both spoken and signed), as she had extremely limited expressive language. Her mother reported occasional spontaneous non-word vocalizations and an occasionally recognizable spoken “mama”, as well as occasional spontaneous signs for “want” and “more”. Her occupational therapist suggested the possibility of a co-existing developmental disorder in addition to hearing differences. However, parents reported that when they had brought up concerns with other members of the treatment team, they had been reassured that she had “lots of strengths,” and had been informed by her audiologist that she was “hearing at 35 decibels with her cochlear implants” (unaided/aided audiograms were requested but ultimately not able to be provided for independent review).

**Exam**

Head, ears, eyes, nose, and throat examination were all unremarkable. Limited eye contact and social engagement was noted for the duration of the visit. She demonstrated persistent repetitive motor and verbal behaviors including constantly circling the room, spinning in a circle, beating her arms against her sides, and flapping her hands. She was repeatedly screeching loudly and saying, “ah ah ah!” In addition, she appeared to be fixated on objects (e.g., pen, ID badge), and enjoyed scribbling repeatedly with a pen. She did not demonstrate any formal expressive language, in American Sign Language or spoken language, throughout the visit.

**Assessment and Plan**

Due to her clinical presentation, a co-occurring developmental disorder was suspected in addition to hearing status. The patient was referred to our hospital’s autism and developmental disorders clinic for further evaluation by a team including a licensed psychologist and a speech language pathologist with a Certificate of Clinical Competence, who also reviewed her available previous testing. Assessment included, but was not limited to, Developmental Profile 3rd edition (DP-3), Brief Observation of Symptoms of Autism—Minimally Verbal (BOSA-MV, which was administered in lieu of an Autism Diagnostic Observation Scale due to COVID restrictions), Childhood Autism Rating Scale 2nd edition standard version (CARS2-ST), Adaptive Behavioral Assessment System 3rd edition (ABAS-3), and Early Classroom Assessment Scoring System (Early CLASS) Functional Communication Assessment. She was ultimately identified with autism spectrum disorder as well as language disorder. Multiple resources were provided, and recommendations were made, including social skills intervention, continuation of speech and occupational therapy, consideration of Augmentative and Assistive Communications technology, consideration of neurodevelopmental evaluation, recommendations for educational programming, and recommendation for Applied Behavioral Analysis (ABA) therapy.

At subsequent follow-up visits, the patient’s mother reported that ABA had been a helpful intervention for her daughter, to the extent that they chose to enroll her in a full-time ABA program rather than in a more traditional self-enclosed or accommodated education classroom. The family continued therapy services, but also elected to find a different Auditory Verbal speech therapist who had more experience in working with children with autism and was able to collaborate with her ABA program. Her parents reported the new therapist was a better fit for the family as well as the patient. The patient’s mother shared with us at subsequent follow-up visits that although the initial discussion around the possibility of autism had been very hard, it was also a relief to get confirmation that something else was contributing to their daughter’s developmental delay. Additionally, she stated that the diagnosis had changed how her family functioned around her daughter, both in how they attempted to connect with her and the educational decisions they made for her in terms of therapies, school enrollment, and use of amplification.

## Discussion

In their 2022 Sentinel Event Alert, the Joint Commission describes diagnostic overshadowing as "the attribution of symptoms to an existing diagnosis rather than a potential co-morbid condition." Cognitive bias can lead a clinician away from considering other alternatives to presenting symptoms, and toward viewing a patient solely through the lens of their established diagnosis. Speed, stress, and lack of training all contribute to an increased risk of inappropriately assigning presenting symptoms to a pre-existing condition (Ospina et al., 2019, p.1).

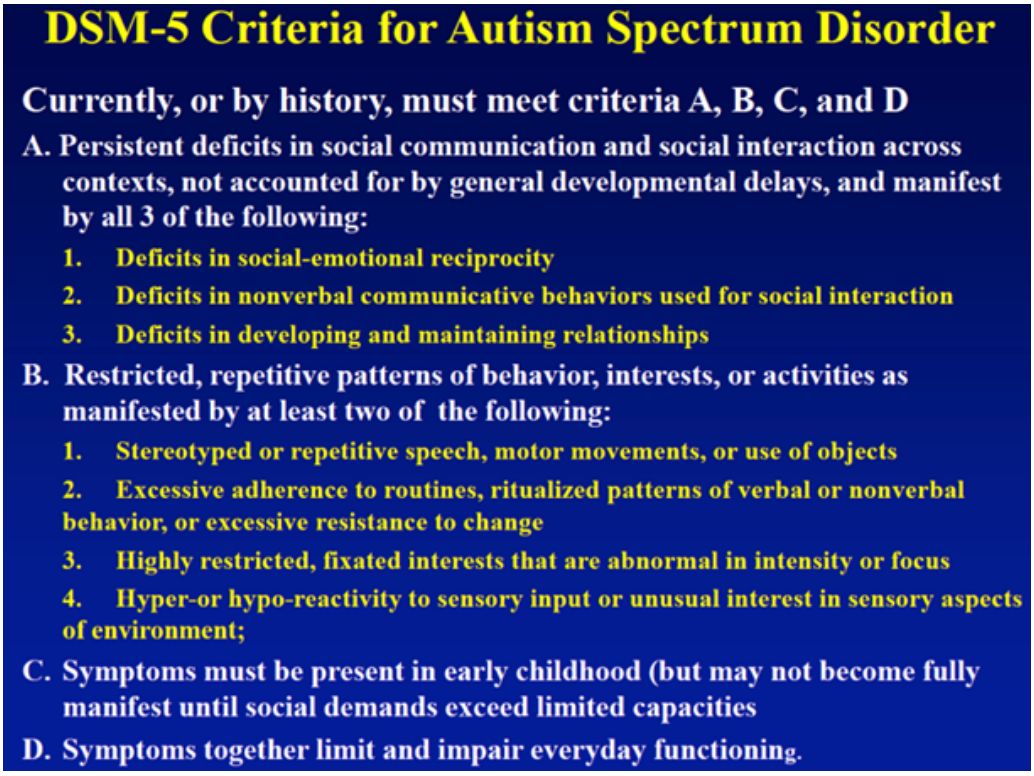
In this case, the patient's deafness appeared to steer most of her clinicians and therapists away from appreciating her additional underlying developmental disorder, potentially due, in part, to viewing her through a *specialty lens* and underappreciating her overarching presentation. This issue can be exacerbated further by the fact that professional efforts to take the time to collaborate and communicate across specialties and disciplines are not traditionally rewarded in the current U.S. medical system, which tends to prioritize productivity, efficiency, and high volume workflow. Professionals may not feel they have the *bandwidth*, depending on their practice environment, to consider involving interdisciplinary specialists when they are not seeing the outcomes they expect as there is often little protected time for this. Consequently, they may continue down the same clinical track with the hope that more time and continuation of therapy may improve outcomes. It is very difficult for one specialist to be all things to all patients and increasing patient complexity warrants increasing diversity of both evaluation and therapy, as well as making system changes that allow for more multidisciplinary collaboration.

Atypical sensory responsiveness is one of the salient diagnostic features of autism, and children with autism who are deaf or hard of hearing can have difficulty tolerating hearing technologies such as hearing aids or cochlear implants (Ludwig et al., 2022; Beers et al., 2014). This may be due to an intolerance for the physical feel of the appliance on or around the ear, the actual sound input, or both. This patient's language development delay may have been mistakenly attributed to implant non-use before further interdisciplinary evaluation revealed an underlying diagnosis of autism that resulted in diminishing sensory tolerance to implants.

In some ways, it is understandable that a clinician unfamiliar with how children who are deaf and not neurotypical present may attribute certain behaviors to "being deaf," particularly if the provider is not familiar with sign language and which movements constitute true language as opposed to repetitive behaviors. Yet, with the current prevalence of autism in the United States estimated at 1:36 children (Maenner et al., 2023), many clinicians have a baseline familiarity with characteristics of autism in children who are hearing (see Figure 1). At least 40–50% of children who are deaf or hard of hearing have one or more coexisting medical/developmental conditions (Bowen & Probst, 2023), which makes clinical evaluation more complex. More specifically, studies indicate an overall increased prevalence of autism in children who are deaf or hard of hearing at 7–9% (Do et al., 2017; Kancherla et al., 2013; Van Naarden Braun, et al., 2015). If this child had been hearing and had presented with lack of eye contact, social avoidance, repetitive behaviors, preoccupation with objects, and lack of expressive language development, the suspicion for a developmental disorder may have been raised sooner due to relatively increased diagnostic familiarity.

**Figure 1**

*DSM-5 Criteria for Autism Spectrum Disorder*




*Note*. Reprinted from Open-Access Journal, *Inquiries*. Singh, A. N. (2014). Increases in the prevalence of autism disorder: Exploring biological and socio-environmental factors. *Inquiries Journal/Student Pulse, 6*(09). http://www.inquiriesjournal.com/a?id=913

This patient's limited access to auditory input stemming from deafness prior to implantation, limited cochlear implant adherence, and delays in spoken language development contributed to diagnostic overshadowing between deafness and autism. To add to the complexity, delays in spoken language development related to being deaf can present with features that resemble those seen in autism: not responding to name, atypical social engagement, limited communication, and so forth. However, children who are deaf and neurotypical, even with delayed linguistic development, demonstrate some marked differences from children who are deaf with autism (Ludwig et al., 2022). Children who are deaf and neurotypical are rarely avoidant of eye contact as they are visual learners, and will make significant use of joint attention to ensure understanding in a conversation (whether using spoken or sign language). They also tend to respond well when provided with adequate access to formal language (sign, spoken, or both), while a child with severe autism may not progress with language development despite supported

**Table 1**

*ASD Diagnosis Considerations in Children who are Neurotypical and Children who are DHH*

**Symptoms of Autism Spectrum Disorder**

- Deficits in social-emotional reciprocity
- Deficits in non-verbal communication in social interactions
- Deficits in relationship building
- Stereotyped or repetitive movements, speech, use of objects
- Inflexible adherence in routines or ritualized behavior
- Fixated interests
- Hyper or hypo-reactivity to sensory input

| Child who is Neurotypical and DHH | Child who has ASD and is DHH |
|---|---|
| • Appropriate social smile<br>• Appropriate eye contact<br>• Engages with others verbally and non-verbally<br>• Can imitate behaviors<br>• Appropriate joint attention<br>• Builds relationships when provided with communication means<br>• May use more gestures/classifiers if exposed to ASL<br>• Shows varied play and interests<br>• Is flexible and can transition without major difficulty | • Social emotional reciprocity and language delays are lower than what is expected for child with hearing loss<br>• Poor eye contact<br>• Reciprocal conversation difficult<br>• Atypical social approach<br>• Poor joint attention, lack of pointing<br>• Difficulty building relationships<br>• Does not respond to name or attention-getting movements<br>• Difficulty understanding others' needs and social cues, including signed emotional cues<br>• Pronoun reversal<br>• Failure to initiate or respond to peers in communication or make/sustain friendships<br>• Language acquisition delays<br>• Difficulty recognizing Deaf culture norms<br>• Shows reduced shared enjoyment<br>• Delayed acquisition of symbolic play skills inconsistent with non-verbal IQ<br>• May engage in echolalia through sign and palm rotation errors (Shield et al., 2017; Shield & Meier, 2012)<br>• Idiosyncratic and made up gestures despite formal sign being taught<br>• Rocking, twirling, flapping, spinning<br>• Highly repetitive play<br>• Resistance to change and difficulty in shifting from preferred interest<br>• Highly specific interests that are atypical in topic<br>• Sensitive to sounds or resistant to wearing hearing aids or cochlear implants |

**Considerations to Avoid Diagnosis Overshadowing**

- Could the social reciprocity and conversation engagement be impacted by language skills of child who is DHH?
- Does the language modality of gestures and vocalization match child's communication partner?
- Does child use eye contact and facial expressions as nonverbal communication (typically a strength for children who are DHH)?
- Does child make eye contact with their interpreter if one is being used?
- Is repetitive speech a symptom of ASD or reflective of receptive language deficits because language development was impacted?
- Is difficulty with change a result of not understanding what is happening (DHH) or inflexible behavior (ASD)?
- Are hearing aids and cochlear implants being monitored by audiologist to ensure proper fit and function, thereby ruling out sensitivity that is not indicative of ASD?

*Note.* Lugwig et al. (2022) provides a detailed comparison of children who are neurotypical and DHH versus children who are DHH and present with ASD. ASD = Autism Spectrum Disorder; DHH = deaf or hard of hearing.

access to it, regardless of language mode(s) provided. (A significant red flag in this particular case was that this child was consistently being presented with two communication modalities, spoken and visual, and was not developing expressive language in either.) Facial expression will often be reduced and restricted compared with children who are deaf and neurotypical. Language development may also have atypical features such as sign copying (sign echolalia) or abnormal hand position when making signs. Repetitive and restricted patterns of behavior, often a hallmark of autism, are not usually a presenting feature in children who are deaf and neurotypical. Table 1 outlines key presenting differences between children who are neurotypical and deaf or hard of hearing (DHH) and those who have Austism Spectrum Disorder (ASD) and are DHH. The child's hearing change in this case study was diagnosed at age 14 months, so this case seems more consistent with diagnostic overshadowing rather than unidentified hearing status presenting similarly to autism.

## Conclusion

Diagnostic overshadowing is a challenge for many clinicians, which can be addressed in part by gaining understanding of how the intersectionality of multiple characteristics and conditions in a single individual can collectively impact their functioning. The need for interdisciplinary input and evaluation is critical in identifying and viewing these children in the context of their whole person, and not just according to a particular specialty or discipline. Children who are deaf with autism spectrum disorder can present a unique diagnostic challenge due to lack of familiarity with how children who are deaf and neurotypical function in comparison, as well as overlapping symptomatology. Providers who recognize clinical features of both children with autism as well as those of neurotypical children who are deaf will be better poised to recognize and support a child who is deaf with autism.

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