# Dually Diagnosed: A Retrospective Study of the Process of Diagnosing Autism Spectrum Disorders in Children Who Are Deaf and Hard of Hearing

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## ABSTRACT

Utilizing a retrospective chart review of 30 children who have been dually diagnosed with hearing loss and autism spectrum disorders (ASDs), this study explores the process of arriving at the diagnosis of ASD in this population. Factors of interest include the age of ASD diagnosis in children who are deaf and hard of hearing, the types of professionals involved in making the diagnosis, and the measures used for assessment. Complications in the diagnostic process are highlighted.

**KEYWORDS:** ASD, autism, deaf and hard of hearing, diagnosis, retrospective study

**Learning Outcomes:** As a result of this activity, the reader will be able to (1) discuss a minimum of two complicating factors in diagnosing autism spectrum disorders (ASD) in children who are deaf or hard of hearing, and (2) discuss the age of diagnosis of ASD in children with hearing loss on the basis of the findings of this retrospective chart review study.



Autism spectrum disorders (ASDs) are complex neurodevelopmental disorders that lead to impairments in social skills and communication. They are typically defined in terms of a "triad of deficits in social reciprocity, communication and repetitive behaviors or interests, each of which can occur at different levels of severity."[1(p.11)] Although there are many reasons to believe that ASD is a neurobiological disorder with a strong genetic

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component, a specific biological marker has not yet been found, leaving behavioral assessments as the only available means of diagnosis. In this context, precise diagnosis in young children is difficult due to "the considerable clinical variation and etiological heterogeneity of ASD."[2] Diagnostic decisions are further complicated by the high incidence of accompanying disorders (e.g., intellectual disability, seizure disorder, fragile X syndrome).[2]

Currently, there is a dearth of information regarding the process of diagnosing ASD in children who are deaf or hard of hearing. The present study adds to the existing knowledge of the process of arriving at the diagnosis of ASD in this population based on a retrospective chart review of children who have been dually diagnosed.

## CURRENT PRACTICES IN DIAGNOSING ASD

### Gold Standard

The current gold standard instruments used for the diagnosis of autism for research rely on clinical evaluation, Autism Diagnostic Interview (ADI-R) or Autism Diagnostic Observation Schedule (ADOS) in an effort to standardize phenotypic criteria across clinical sites.[3] Results of a recent extensive review of all current diagnostic tools found that ADI-R in combination with the ADOS have "the greatest evidence base and highest sensitivity and specificity."[3(p.329)] In clinical research studies, accuracy rates when utilizing the ADI-R and ADOS combination was 80.8%.[3]

However, the ADOS and ADI-R are lengthy assessments. There is a pressing need to develop a similar level of resources for cost-effective and rapid phenotyping both for research purposes and clinical use.[3] These efforts continue especially in the light of the developments introduced by the *Diagnostic and Statistical Manual of Mental Disorders,* Fifth Edition (DSM-5).[4] Furthermore, best practices in ASD diagnosis indicate the need for a comprehensive evaluation with a multidisciplinary team, including a psychologist, neurologist, and a developmental pediatrician. Psychiatrists, speech therapists, and other professionals can also offer important insights in the diagnosis of children with ASD.

### Diagnosis of ASD in Special Populations

As challenging as a diagnosis of autism in an individual without accompanying disorders can be, such a diagnosis becomes extremely challenging in cases where a dual diagnosis of autism and another disorder is warranted. Often autistic-like behaviors are mistaken for behaviors resulting, for example, from loss of vision or hearing. If a child is first recognized as having a hearing or visual loss before having an autism diagnosis, some of the autism-related behaviors may be mistaken by parents and professionals as a consequence of that sensory loss alone.

Despite the challenges involved, researchers and clinicians have identified ASD in various special populations. For example, it was previously believed that Down syndrome and ASD could not coexist.[5] Parents were informed of their child's cognitive limitations and were informed that atypical behaviors that might arise were associated with Down syndrome, without further investigation into potential other contributing factors. Yet, it has been documented that the diagnosis of Down syndrome in combination with ASD "represents a distinctive set of aberrant behaviors marked by characteristic odd/bizarre stereotypic behavior, anxiety, and social withdrawal."[3(p.87)]

Another example would be Williams syndrome, a disorder noted for individuals with their high sociability and empathy for others.[6,7] As a hallmark of this syndrome, "individuals are typically fearless and engage eagerly and often impulsively in social interaction, even with strangers."[8] This characteristic often leads researchers and clinicians to a consideration of Williams syndrome as a type of "anti-autism"—essentially clinical opposites. Yet, such a theory does not hold up to scrutiny. The two disorders coexist more frequently than previously believed.[9]

### Confounds of ASD and Hearing Loss

The literature in the field of autism and hearing loss indicates that it is difficult to make a diagnosis of ASD in the presence of hearing

loss. To date, the accumulated knowledge in this area regarding the process of diagnosis in this population has primarily been based on small case studies. Additional information has been derived from clinicians' conceptualization of the overlaps, similarities, and differences in the presentation of children with ASD, hearing loss, and combined ASD + hearing loss. Regarding the latter group, although this is a topic of increasing interest, relatively little is known at this time.

## THE PRESENT STUDY

This research involved a retrospective chart review of the medical records of children diagnosed as deaf or hard of hearing (bilateral; with any degree of hearing loss) and an ASD (now known as ASD, but documented in the medical records as PDD NOS (pervasive developmental disorder not otherwise specified), Asperger syndrome, or autism. This research was conducted at a large academic hospital for children. The records examined dated from 2005 to early 2014. The authors are clinician/researchers who have considerable experience with neurodevelopmental disorders in children. Two researchers have expertise in working with children who are deaf or hard of hearing; the other has expertise in working with individuals with ASD.

## METHODS

The present study utilized a retrospective chart review to explore the process of diagnosing ASD in children who are deaf or hard of hearing. Methodological considerations were implemented in conducting retrospective chart review studies incorporating current best practices for such studies.[10,11] Gearing and colleagues proposed nine steps be involved in acquiring the data for a retrospective chart review.[11] The steps taken in the present research are detailed below.

### Research Formulation

The following research questions and hypotheses drove this investigation:

Hypothesis 1: If ASD is prevalent among children in the general population, then it should also be present in the population of children who are deaf or hard of hearing.

Hypothesis 2: If, in the best of cases ASD is challenging to diagnose consistently and accurately in children with normal hearing by professionals trained in neurodevelopmental disorders,[12] then it is likely that diagnosing of ASD in children who are deaf or hard of hearing would be complicated further. We would expect that arriving at a diagnosis would be problematic and that the testing done would be incomplete.

Hypothesis 3: If deafness or hearing loss in early childhood is a factor that can contribute to language delays, then we hypothesize that when language delays are present in young children who are deaf or hard of hearing, family members and professionals may attribute them to the child's hearing status (i.e., the hearing loss will be "blamed" for language delays), which could result in later diagnosis of ASD in children with hearing loss relative to hearing peers.

#### LITERATURE REVIEW

The authors conducted an extensive review of the literature available on ASD and deafness and hearing loss. This is an area of investigation that has not yet been well established; thus, the research on this topic is rather limited.

#### PROPOSAL DEVELOPMENT AND ETHICS

The authors familiarized themselves with the health records, inspecting several charts, and examined where the information relevant to the present study could be found. They have worked with these records extensively, as well, in their roles as clinicians. This research was approved by the institutional review board of the hospital at which this project was completed.

#### DEVELOP DATA ABSTRACTION INSTRUMENT AND GUIDELINES FOR ABSTRACTION

A record review protocol was developed, tested, revised, retested, and then utilized to gather data in a uniform and consistent manner. The authors established a coding strategy and discussed the variables to be captured.

### PILOT STUDY

The researchers conducted a pilot study to determine the feasibility of the investigation. After a discussion of the findings from the pilot study, adjustments were made for the larger study.

### DATA ABSTRACTION

The data were collected by the authors who, as noted above, have both research and clinical experience. The authors discussed conflicting or confusing information found in the medical charts and achieved consensus about how to capture information when questions arose.

## Participants

Medical records were reviewed for 30 patients who had a dual diagnosis of sensorineural hearing loss and ASD. More than half (53%) of the children identified with ASD and hearing loss had been diagnosed at other facilities and were then referred to this academic medical center. Thus, the content of the medical records varied, depending on the extent to which earlier records could be accessed. A total of 10 additional records were examined yet not included in this analysis, as they lacked the minimum amount of information needed to be informative in this study. See Table 1 for demographic information of the participants.

Degree of hearing loss varied, and hearing status in each ear was often heterogeneous. Overall, however, the majority of patients had severe, severe to profound, or profound hearing loss. Significantly more boys than girls had been given the dual diagnosis. Communication modality also varied—roughly one-third of the parents described their children as users of a signed language (most often American Sign Language, but some reported "gestural communication");

**Table 1 Demographic Information of Study Participants**

| | | Subjects Meeting the Criteria |
|---|---|---|
| Diagnosis Given | | |
| | Other facilities | 53% (16) |
| | This facility (various departments) | 46.67% (14) |
| Gender | | |
| | Male | 70% (21) |
| | Female | 30% (9) |
| Degree of Hearing Loss | | |
| | Severe, severe–profound, profound | 70% (21) |
| | Mild–moderate, moderate | 26.67% (8) |
| | Variable | 0.03% (1) |
| Communication Modality | | |
| | ASL or gestural communication (ASL not well developed) | 36.66% (11) |
| | Spoken language (English, Spanish, Portuguese, Greek) | 33.33% (10) |
| | Combination – ASL or gestural communication + Spoken Language | 30% (9) |
| Hearing Status of Parents | | |
| | Deaf or hard of hearing | 33.33% (10) |
| | Hearing | 66.67% (20) |
| Use of Assistive Listening Devices | | |
| | Hearing aids* | 36.66% (11) |
| | Cochlear implants* | 33.33% (10) |
| | None | 30% (9) |

Abbreviation: ASL, American Sign Language.
*Assistive listening devices have been used. Several these children do not use them consistently, show resistance to using them, or have stopped using them for periods of time or altogether. These data only show the number of children for whom hearing aids or cochlear implants have been utilized.

one-third indicated that their children were oral communicators; and one-third reported that their children used spoken language in combination with signs. Of the 30 children who were dually diagnosed, 33% had at least one deaf parent. Use of assistive listening devices by this group of children was common, with 70% reported using of hearing aids or cochlear implants to access auditory information.

Many of the children dually diagnosed with ASD and deafness or hearing loss were also found to have additional medical, behavioral, or psychiatric diagnoses. Although level of intelligence was not documented for many of the subjects, when cognitive testing results were found in the medical record, intellectual abilities in this group tended to be reduced relative to the general population.

## RESULTS

Consistent with hypothesis 1, we found that ASD is represented in the deaf and hard of hearing population. Hypothesis 2 predicted that the diagnosis of ASD in this population would be complicated, that arriving at the diagnosis would be challenging, and that the process (and the records documenting that process) would be incomplete. This hypothesis was supported. Furthermore, hypothesis 3 proposed that the diagnosis of ASD in children with hearing loss would be complicated by the misattribution of symptoms to the hearing loss, resulting in later diagnosis of ASD than is typical in the general population[13]; this hypothesis, too, was supported.

Several interesting facts and themes emerged from the review of the records. This information adds to the understanding of the process of arriving at a diagnosis of ASD in children with an identified hearing loss.

### Parent Driven

Medical records indicated that many of the professionals involved in the early care of the patients documented being perplexed by the overall presentation of the child. These care professionals often described features consistent with the diagnosis of ASD in their clinical notes, yet did not document taking the steps necessary to arrive at a diagnosis of the condition. For a surprising number of children (60%), the parents/caregivers sought numerous opinions and arranged for additional consultations with professionals to arrive at a diagnosis of ASD. Based on review of the clinical record, it was apparent that many professionals were hesitant to use the ASD label. It was also clear from the records that, in many cases, the parents were aware that the child had features of ASD and were seeking confirmation from professionals. Consistent with the literature,[14] we found that parents' initial concerns frequently centered on communication. Communication delays and idiosyncrasies were often attributed, by parents and professionals alike, to the hearing loss and resultant inconsistent or lack of access to auditory information. Many parents, however, come to perceive the communication challenges, in combination with behavioral manifestations, as beyond what could be attributed to hearing status alone, and sought confirmation of their suspicions/concerns for their children.

### Later Diagnosis

The average age of diagnosis of ASD in children with typical hearing is just over 3 years.[13] In this clinical sample of 30 children with hearing loss who were diagnosed with ASD, the average age of the preliminary diagnosis was 4.5 years. In nearly all cases, a confirmatory diagnosis was given after a working diagnosis of ASD had been in place for some time. The average age at which a formal diagnosis of ASD was given was 6 years, 4 months (over the 9-year span from which charts were reviewed).

In recent years (charts from 2011 to 2014), several children were diagnosed at younger ages. In this subset, four of the children who were deaf and were diagnosed earlier (i.e., before 3 years of age) had older siblings who had already been diagnosed with ASD. It is known that when older siblings have been diagnosed with ASD, younger siblings are more likely to be diagnosed at earlier ages.[11,15] Additionally, one of the children was diagnosed with ASD significantly later in life, at age 18. In this particular case, diagnosis was impacted by the severity of the individual's medical complications. Excluding these five individuals from the analysis, the average age of initial diagnosis was 4.8 years

and the mean for confirmatory diagnosis was 6.25 years for the population reviewed.

Of course, it is not possible to ascertain whether earlier diagnosis in recent years is solely attributable to sibling status and is therefore an anomaly or if perhaps earlier diagnosis may be an emerging trend. As will be discussed, a central recommendation of this article is the need for early dual diagnosis for this population. See Table 2 for details.

## Diagnosis Given by a Variety of Professionals

The initial diagnosis of ASD and the subsequent confirmation of ASD were completed by professionals from a variety of backgrounds. Often, the initial diagnosis or suspicions of a diagnosis were raised by people outside of a medical setting, whereas confirmatory diagnosis was more often achieved by professionals in the hospital for this sample. The professionals credited with diagnosing ASD are listed here as they were reported by the parents of the children in this study. In some cases, the diagnostician was unknown because the medical record indicated that the child "had been diagnosed" and no further information about who had given the diagnosis nor how it had been determined was available. Table 3 provides details.

## Ineffectiveness of Assessment Using Current Measures—Diagnosis by Observation

At present, the gold standard for diagnosis of ASD for research purposes is administration of the ADOS in combination with the ADI-R (see discussion in Lee et al).[3] It is important to note that, clinically, this standard is often not achieved. Although many clinical teams recognize and appreciate the value of input from standardized measures of ASD, administration can be time-consuming and expensive. In practice, the diagnosis of ASD is often dependent upon formally administered diagnostic measures; clinical observation in combination with parental support tend to be crucial in determining the diagnosis.[16]

In this sample of children with hearing loss, diagnosis of ASD was made largely on the basis of clinical presentation. The ADOS was attempted with two of the children; the ADI-R was used in two different instances. Although occasionally efforts were made to use these measures, professionals documented that utilization of these measures was not deemed to be relevant or valid for these patients. Particularly in light of communication challenges, administration of ASD diagnostic measures was perceived as unreliable. The most frequently used means of arriving at an ASD diagnosis included nonstandardized observation of the child in combination with parent interview; this occurred in the vast majority of the cases. Several other measures were used in the process of diagnosing ASD. These are highlighted in Table 4.

**Table 2 Age of Diagnosis of Autism Spectrum Disorder in Children Who are Deaf and Hard of Hearing**

| | Age | | | |
|---|---|---|---|---|
| | Range | Mean (y) | Median (y) | Mode (y) |
| Initial or provisional diagnosis | 16 mo –18 y | 4.5 | 3.6 | 3.6 |
| Subsequent or confirmatory diagnosis | 22 mo –18 y | 6.35 | 5 | 5.6 |

**Table 3 Professionals Credited with Diagnosing Autism Spectrum Disorder in Children Who Are Deaf and Hard of Hearing**

| | Percentage involved in diagnosing* |
|---|---|
| Developmental pediatrician/ developmental medicine | 23.3% (7) |
| Psychiatrist | 20.0% (6) |
| Neurologist | 20.0% (6) |
| Psychologist | 16.6% (5) |
| Pediatrician | 10.0% (3) |
| Neuropsychologist | 03.3% (1) |
| School psychologist | 03.3% (1) |
| Early intervention service provider | 03.3% (1) |
| Unknown | 10.0% (3) |

*Percentage exceeds 100% because some children were assessed by multi-disciplinary teams consisting of more than one professional.

**Table 4 Diagnostic Tools Used in the Diagnosis of Autism Spectrum Disorder in Children Who Are Deaf or Hard of Hearing**

| | |
|---|---|
| Observation + parent report | Bayley Scales of Infant and Toddler Development, 2nd edition |
| Autism Diagnostic Observation Schedule | Brain Electrical Activity Mapping (BEAM study) |
| Autism Diagnostic Interview–Revised | Vineland Adaptive Behavior Scale, 2nd edition (Vineland-II) |
| Behavior Assessment System for Children, 2nd edition | Clinical Evaluation of Language Fundamentals, 4th edition–Pragmatics Profile |
| Scales of Independent Behavior–Revised | Social Skill Rating System |
| Childhood Autism Rating Scale | Neuropsychological Battery |

## DISCUSSION

For professionals already working with children who are deaf or hard of hearing or involved in diagnosing ASD, we urge greater awareness of the possibility of the dual diagnosis. Results, as noted previously, indicated individual differences on the part of professionals with respect to comfort in giving the dual diagnosis. It will also be important that clinicians understand the ways in which ASD presents in children who are deaf or hard of hearing in order to understand the intricacies of the dual diagnosis. This in turn suggests that additional training may be warranted on the part of diagnosticians.

The most frequent diagnosis in this clinical sample was PDD NOS. It seems that PDD NOS was often used as a cautionary hedge. Diagnosticians documented being hesitant to label autism with certainty and regularly indicated that the diagnosis should be considered provisional or speculative. Under DSM-5, such a diagnosis will become problematic.

The special nature of the dual diagnosis puts children at risk with respect to decreased language access (visual or spoken), which is essential for advancement across numerous domains, including not only linguistic development, but also cognitive, social, and behavioral development.

Naturally, the later the diagnosis, the later the interventions and supports are put into place. This has myriad consequences, including lost opportunities for building a solid language foundation and reduced chances for building constructive, positive, social connections, as well as generally reduced cognitive development and increased behavioral manifestations. Earlier diagnosis can help to reduce these risks.

Optimal treatments for children with this dual diagnosis are not simply the union of standard practices in supports offered by either condition. Rather, the diagnoses demand that treatment approaches be tailored and, perhaps, call for the development of unique treatment protocols.

### Limitations of the Research

There are several limitations to the present research. Due to the relatively low incidence of this dual diagnosis, the sample size is limited. The inclusion of additional research participants would yield more robust findings. The study was further limited by the variable information found in the medical records; some records were incomplete. Particularly when patients had transferred care from other hospitals to this site, information about what happened early in each child's life was often not as comprehensive as desired. In several of the records, warning signs of ASD were documented. Then, at a future date, the records indicated that the child had been diagnosed on the spectrum. It was not always clear from the medical record precisely when the diagnosis of ASD had been given; in some cases, there was not documentation available to show which professional(s) had labeled the child nor what instruments they used to arrive at the diagnosis.

### Future Directions

As documented elsewhere (see Mood and Shield, in this issue),[17] currently available assessment instruments do not adequately capture ASD in this population. The present study

offers further evidence that current measures for diagnosing ASD in the general population have not frequently been used. Standardized ASD instruments have not yet been sufficiently adaptable for use during assessments with children who are deaf of hard of hearing. There is a need for instruments that can be properly administered and can capture the essence of the clinical presentation. Reconceptualizing and standardizing of current measures by professionals with expertise in both ASD and hearing loss is needed. Establishing new measures that can be validated and that would allow accurate and efficient phenotyping made also be necessary.

Larger investigations, involving analysis of more dually diagnosed children, can further help to inform the field about this. Studies that incorporate larger numbers will allow for a more thorough and in-depth review of the various other factors that may contribute to the diagnostic process (e.g., IQ scores, reports from families, records from other medical centers, and so on). With this information, additional themes, major trends, and correlations in the diagnosis of ASD in children who are deaf or hard of hearing may become more evident.

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