DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY SCOPING REVIEW

Editor's Choice

# Communication in Angelman syndrome: a scoping review

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**ABBREVIATIONS**

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| AAC | Augmentative and alternative communication |
| ENG | Enhanced natural gesture |

**AIM** A scoping review was conducted to examine and evaluate empirical data on the communication profile of Angelman syndrome beyond the described dissociation between receptive language and speech.

**METHOD** Three databases (PsycINFO, Embase, and Web of Science) were searched to retrieve articles investigating communication in Angelman syndrome. Seventeen articles investigating the broader communication profile were found; their methodology was evaluated against quality criteria.

**RESULTS** Despite the absence of speech, individuals with Angelman syndrome have a wide repertoire of non-verbal communicative behaviours, mainly characterized by gestures, although advanced forms such as symbolic communication are used by some individuals. The use of communicative forms differs between the genetic aetiologies of Angelman syndrome; individuals with non-deletion aetiologies typically have greater communicative abilities.

**INTERPRETATION** The broader communication profile of Angelman syndrome is characterized by diverse and multimodal abilities, including some use of symbolic forms of communication that appears atypical given the absence of speech. This is suggestive of a probable dissociation between speech and other expressive forms of communication, indicating an isolated speech production impairment. This highlights a need in this population for alternative communication and specific input from services tailored to support the nuances of the communication profile of Angelman syndrome.

Expressive language delay and communication impairments are characteristic of individuals with intellectual disabilities, with variability in the trajectory of acquisition and resultant profile across domains of communication.[1,2] A number of factors are associated with expressive language growth in individuals with intellectual disabilities, including vocabulary comprehension, prelinguistic communication, and parental response contingency.[1,3,4] These factors can account for the variability seen in expressive language development in individuals with intellectual disabilities and can highlight key areas that could be targeted for early intervention to improve communicative abilities.

In intellectual disability research, there is frequent imprecision in the use of terms such as language and speech, despite the significant difference between language and speech impairments. In populations where communication and language problems are common, such as individuals with intellectual disabilities, there is a need for specificity and clarity of these terms and hence concepts to tailor support and intervention appropriately.

Differences in strengths and weaknesses across genetic syndromes with regard to different communicative components highlight this need for specificity. For example, Singer Harris et al.[5] compared the development of communication in infants with Down and Williams syndromes. Their findings showed different communication profiles, with infants with Down syndrome having a significant gestural advantage, whereas infants with Williams syndrome had greater strengths in grammar. Interestingly, both syndrome groups were equally delayed by approximately 2 years in their expressive language acquisition; furthermore, there was no difference in the total number of words spoken between the two groups. This highlights both the theoretical importance of recognizing genetic aetiologies associated with communication difficulties, in terms of understanding the roles gene disorders may play in language and communication, and the clinical importance of tailoring specific and effective interventions, possibly at the syndrome level.[6]

One syndrome associated with intellectual disabilities and where expressive language and communication difficulties are a key characteristic is Angelman syndrome. Angelman syndrome is a rare genetic disorder caused by disruption to genetic information on the maternal copy of chromosome 15q11-13, specifically the *UBE3A* gene. It has an estimated prevalence of between 1 in 12 000 and 1

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in 20 000 live births.[7–9] Four different genetic aetiologies of Angelman syndrome have been identified, with the most common being the deletion of *UBE3A*, which occurs in between 65% and 75% of individuals; 5% to 11% of individuals have Angelman syndrome due to a mutation of *UBE3A*; paternal uniparental disomy of chromosome 15 occurs in around 3% to 7% of individuals; and 3% to 5% of individuals with Angelman syndrome have an imprinting defect.[10,11]

The clinical features of Angelman syndrome are severe-to-profound intellectual disabilities, ataxic gait, and minimal or absent speech.[12] Additional behavioural characteristics highly associated with the syndrome include frequent laughing and smiling, high levels of sociability, feeding problems, sleep disturbance as well as hyperactivity, and a short attention span.[10,13,14] Genotype–phenotype correlations have been reported in Angelman syndrome, with individuals who have non-deletion aetiologies presenting a 'milder' clinical and behavioural phenotype.[15–18]

Lack of speech is notable in Angelman syndrome, with between 71% and 90% of individuals never or rarely producing any speech.[19,20] In those individuals who have speech, vocabulary is limited, with the literature reporting a spoken vocabulary size of between two and 15 words, with an average of five words.[21–24] However, reportedly these words are rarely used for communicative purposes.[25] Furthermore, some studies have suggested that the level of intellectual disability cannot account for the absence of speech[26,27] because the receptive language abilities of some individuals would typically be associated with the presence of expressive language.[27]

Studies investigating the phenotypic differences between genotypes in general adaptive skills, including communication, have highlighted that individuals with non-deletion aetiologies have significantly greater receptive language skills compared to individuals with a deletion. In contrast, the markedly reduced level of expressive language appears homogenous across the genetic aetiologies. This suggests that *UBE3A* may be implicated in speech production[16,18] and further strengthens the argument that minimal or absent speech may not be the result of receptive skills or global intellectual disability.

Despite the homogeneity and atypicality of absent speech in Angelman syndrome, in addition to the established discrepancy between receptive and expressive language, there is limited research quantifying the impairment across other domains of communication, such as gestures. Knowledge about the communication profile and specificity of impairments would enable a better understanding of possible targets for alternative communication methods as well as effective intervention. Furthermore, because of the genetic basis of this syndrome, understanding the communication profile of Angelman syndrome has broader implications for understanding the development of communication and speech production specifically. Given the limited literature, scope of the topic of communication, and need to clarify and identify gaps in the current understanding, it is beneficial to conduct a scoping review.[28]

**What this paper adds**

- Although absent speech is near universal, a diverse profile of other communicative abilities has been reported.
- Parental reporting has been predominantly used to assess the communication profile of Angelman syndrome.
- Literature that investigates the specificities and possible dissociations in such a communication profile is limited.

The aim of this scoping review is to search the published literature to summarize the empirical data on communication in Angelman syndrome and evaluate the methodology of these studies. This will enable gaps in research to be highlighted and suggestions for future research in this area to be generated, alongside providing greater clarity of the profile that can be used to inform syndrome-sensitive intervention and assessment.

## METHOD

### Search strategy

A systematic search of three databases (Embase, PsycINFO, and Web of Science) was conducted. The keywords 'communicat*' and 'Angelman syndrome' were combined using the Boolean operator 'AND' to retrieve any articles published between January 1960 and October 2018 (week 4). No a priori protocol was registered, but further information on the process can be obtained from the corresponding author on request. Figure S1 (online supporting information) shows the search process and retrieval of the final articles used in the review.

### Study selection

To be included in the review, studies must have been published in a peer-reviewed journal between January 1960 and October 2018. Only published research was considered to ensure that the validity of the summarized findings had been subjected to peer review. Eligibility criteria required articles to be written in English. Additionally, one of the main aims of the review was to explore the broader communicative abilities of individuals with Angelman syndrome. The scope of this aim needed to extend beyond descriptions of speech and expressive language, and the dissociation with receptive language, since this is already widely acknowledged and forms part of the diagnostic criteria.[12] Case studies were excluded because of sampling bias from focusing on atypical cases and not providing data at a group level. Two reviewers (EP and RR) independently completed the search process and screening procedure to verify the accuracy of the selection of studies.

## RESULTS

### Search results

A total of 17 articles were retrieved, which are presented in Table SI (online supporting information) in the order of the themes discussed within the review. Themes were chosen and agreed on based on the reading of full texts and

the main topic areas investigated in these studies. The final themes chosen were: forms of communication; augmentative and alternative communication (AAC); functions of communication; and communication interventions. Findings from the studies were charted under the relevant theme and were further grouped based on common topics for synthesis; for example, findings related to gesture use were grouped under the theme 'forms of communication'.

The methodological quality of papers was assessed based on amended criteria from Cross and Hare,[29] which were originally created based on reported best practice for behavioural phenotype methodology. Studies were rated from 0 to 2 on six areas: control group; sample size; recruitment; syndrome diagnosis; methodology; and appropriate statistics/comparisons. A total score was obtained for each article (Table SI). Four papers that implemented communication interventions were excluded from this evaluation because the criteria were not appropriate for intervention designs; thus, 13 articles remained for evaluation. Inter-rater reliability was assessed for six included articles, with $\kappa$=0.85; thus, reliability was excellent.

### Forms of communication

Most studies focused on the methods individuals use for expressive communication. There is consensus in the reviewed literature that individuals with Angelman syndrome are multimodal communicators who use a wide variety of communicative methods including non-symbolic, symbolic, and AAC.[19,22,25,30–32] The most commonly used forms of communication reported in the literature come under non-symbolic communication, such as non-speech vocalizations, physical manipulations of others, and gestures.[19,22,25–27,32] These forms of expressive communication are evident regardless of the genetic aetiology of Angelman syndrome.[19]

Gesturing is the most prevalent form of communication in individuals with Angelman syndrome,[19,25,27,32] even when the individual is using more advanced forms of communication such as an AAC device.[29,32] Early research by Jolleff and Ryan[26] stated that only five out of their 11 participants with Angelman syndrome would show gesturing, as reported by their parents using a standardized questionnaire. Further research that has explored different types of gestures shows slightly higher prevalence rates, with estimates for deictic gestures (gestures that establish reference by directing attention or indicating an object or event) between 50% and 100%[24,32,33] and specific deictic gestures, such as pointing, being used by between 40% and 55%[22,32] of individuals with Angelman syndrome; reaching for desired objects is used by 75% of individuals with Angelman syndrome.[22] Representational gestures are not as common as deictic gestures in Angelman syndrome.[24,33]

With regard to the different aetiologies of Angelman syndrome and the use of gestures, Jolleff et al.[34] reported that individuals with a non-deletion aetiology showed more gestural communication than individuals with a deletion. However, the sample size in this study was small, with only seven participants in the non-deletion group. In a more recent study with a larger sample size, there were no significant differences between the aetiologies for the occurrence of gestures, with 83% of individuals with deletion and 76% of individuals with non-deletion mechanisms using gestures.[19] This research only studied the occurrence and not the types of gesture used by individuals, an area where further research is required.

Another form of non-symbolic communication that has been reported in Angelman syndrome is physical manipulation of others.[19,22,26,27] Alvares and Downing[22] found that 50% of parents of a child with Angelman syndrome reported that their child used physical manipulation to communicate. This finding is further corroborated by Calculator[19] in a larger sample of 182 parents, with around 60% of individuals using physical manipulation 'often' or 'very often'. Similarly, around 60% of individuals used non-speech vocalizations to communicate,[19] which have been further detailed as simple vocalizations that are characteristic of typically developing children between 0 and 6 months old.[33] Other non-symbolic forms of communication reported in the literature are body movements, facial expressions, and eye gaze.[25,32]

Although used less frequently than non-symbolic forms, symbolic forms of communication such as signing and speech are used by some individuals with Angelman syndrome.[19,22,25,32] Earlier direct observational research by Jolleff and Ryan[26] and Penner et al.[27] suggested that individuals with Angelman syndrome did not show any forms of symbolic communication. However, more recent research has suggested that the use of symbolic communication is greater than previously described, which may be because of smaller samples, use of direct observation versus parental reporting, and also cohort effects (e.g. institutionalization) in the earlier research. In an online survey of 20 families, 50% reported that their child with Angelman syndrome used some form of signing to communicate, with a further 35% of individuals spontaneously and functionally using signs. The range of signs reported ranged from two to over 200, although only two individuals used over 10 signs.[22] The reasons why these two children had such an extensive repertoire compared to the majority were not explored by the authors. Additionally, parents reported that when their child used signs, they were approximations and hard to understand for those who were unfamiliar with the child. As well as signs, Quinn and Rowland[32] found that other forms of symbolic communication such as concrete symbols, like using a picture (e.g. the picture of a drum to represent music) or an object (e.g. a shoelace to symbolize a shoe), are used by between 18% and 47% of individuals with Angelman syndrome. Abstract symbols, such as a line drawing of a reaching hand to represent help, were also used by 13% to 30% of individuals.

There are also significant differences in the use of symbolic forms of communication between the genetic

aetiologies of Angelman syndrome.[19,25] This contrasts with the lack of differences found for non-symbolic forms. Calculator[19] used parental reporting to explore the differences in forms of communication across individuals with and without deletions. The results highlighted significant differences for the use of enhanced natural gestures (ENGs), signing, and speech, with individuals with non-deletion aetiologies using these methods more frequently. This is further supported by Didden et al.,[25] who reported that individuals with non-deletion aetiologies used signs more frequently than those with a deletion.

Other characteristics of Angelman syndrome alongside genetic aetiology have been shown to be related to the use of symbolic forms among individuals. Didden et al.[25] reported that individuals with severe intellectual disabilities signed more than those with profound intellectual disabilities. In addition, they found that individuals who experienced epilepsy were less likely than those without epilepsy to use symbolic forms of communication. Furthermore, epilepsy was also found to be related to some non-symbolic forms of communication, such as pointing, with individuals with epilepsy being less likely to point. In support of this finding, wider research in the general population has shown that the presence of epilepsy and seizures is related to the presence of language disorders.[35] However, Didden et al.[25] did not consider the level of intellectual disability as a covariate in their analysis. This is an important aspect to consider given that the link between epilepsy severity and level of intellectual disability is well established in other genetic syndromes associated with high rates of epilepsy (e.g. tuberous sclerosis complex).[36,37]

In addition to research reporting on the prevalence of forms of communication, Calculator[31] investigated the perceptions of 174 parents of the importance of a range of communication methods used by their children with Angelman syndrome. Parents ranked non-symbolic forms as the most important forms of communication for children with Angelman syndrome, even when considered against AAC and symbolic forms of communication. In particular, gestures were rated by 84% of parents as very or extremely important, followed by non-speech vocalizations (76% of parents) and physical manipulation (72% of parents). Thus, in his conclusion, Calculator[31] highlighted the importance of supporting an individual's non-symbolic communication across their lifespan.

In general, the literature exploring forms of communication in Angelman syndrome is comprehensive, with a variety of forms explored and described alongside the identification of factors explaining variability in the presence and use of different forms of communication. However, the results focus predominantly on the occurrence of forms based on informant reporting. Future research should focus on using direct observation to assess the wider context of forms of communication and their use to determine the extent of a possible dissociation between other expressive forms of communication and speech in Angelman syndrome. This, in turn, will provide better understanding of how skills may be transferred to and utilized in alternative communication methods.

### Use of AAC

AAC is used by individuals with communication difficulties to aid communication with others by supplementing or replacing spoken language. It comprises a wide variety of methods, from gestures to 'high-tech' devices, such as computer-based systems (for an overview, see Murray and Goldbart).[38]

Aided forms of AAC, both low- and high-tech, are used by between 62% and 70% of individuals with Angelman syndrome.[19,22] Calculator[19,31] reported a wide range of electronic AAC devices used by individuals with Angelman syndrome, with individuals tending to use more than one device to communicate. With recent advances in mobile technology, there has been a shift towards utilizing apps on tablets as a means for individuals to communicate, with 48% of individuals with Angelman syndrome using an AAC app on a tablet as their most advanced AAC device.[31] As well as citing the devices used, Calculator[19] reported differences in the AAC systems used by individuals with a deletion and those with non-deletion aetiologies. Individuals with a deletion aetiology used less advanced AAC systems, such as single-message voice output communication aids, and were less likely to progress on to a more advanced device than individuals with non-deletion aetiologies.

Regardless of the complexity of the device, an important factor to consider is the individual's success with their device. Calculator[31] stated that only 4% of 142 parents reported that their child with Angelman syndrome was totally successful with their most advanced AAC device. However, 70% reported that their child was communicating successfully with their device at some level. Alternatively, 4% reported that their child was totally unsuccessful in using their device. Factors such as support, which may play a role in an individual's success with a device, were not investigated.

An individual's success with a device is dependent on whether they 'accept' the device. Acceptance is defined as the extent to which an individual and their family and/or carers will willingly integrate AAC devices into their daily lives.[39] Calculator[19] reported that around 60% of individuals with Angelman syndrome accept their most advanced AAC device; using thematic analysis of parental reporting, Calculator highlighted a number of factors that influence an individual's acceptance of advanced AAC devices.[30] The most frequently reported factors were the individual's ability to communicate more effectively and purposefully alongside contingent responses from those around them. In contrast, the most cited reason for rejecting the AAC device was the individual's lack of understanding or valuing the device as a communication tool, for example, seeing it as a toy more than a means of communication. Therefore, it was concluded that for an individual to accept a device and use it as an effective form of communication, the

complexity of the device has to match the developmental level of the individual.

Despite advances in mobile technology and AAC devices, non-symbolic forms are still the preferred method of communication by individuals with Angelman syndrome[30,31] as well as the symbolic forms that some individuals choose to use over their AAC device. Furthermore, most parents view AAC devices as important to some degree and value the clarity they add to their child's communication when compared to other forms of communication. However, a smaller proportion of parents view AAC devices as very or extremely important compared to the importance of non-symbolic communication.[31]

This is currently a limited area of research in Angelman syndrome, with the possibility of significant sample overlap in the literature, although there are substantial and representative sample sizes. It is an area of research that needs to be extended, especially given that AAC devices are a viable and necessary intervention to assist with communication in Angelman syndrome. In particular, further research is needed on understanding the acceptance and success of devices alongside devising appropriate learning strategies for individuals.

### Functions of communication

Although explored less than forms of communication in Angelman syndrome, a small number of studies have investigated the functions of communication, such as the use of proto-imperatives (e.g. requesting) and proto-declaratives (e.g. directing another's attention, sharing of experiences). There is a consensus that requesting is the best developed and most widely used function, whereas imitation and commenting appear to be areas of comparative weakness in the communication profile.[25,27,32,40,41] Using parental reporting, Didden et al.[25] explored the widest range of communicative functions in individuals with Angelman syndrome. Ten different functions, and how individuals communicated these functions, were explored. Their conclusions highlighted that individuals will communicate for a variety of reasons, including rejecting and protesting, and requesting and commenting, the latter providing evidence of use of proto-declaratives in Angelman syndrome. Furthermore, requesting information and imitation were the least developed areas of communication.

These findings were further corroborated in 300 individuals with Angelman syndrome. Quinn and Rowland[32] used a data mining strategy to explore which communicative strategies individuals with Angelman syndrome use to refuse, obtain, interact socially, and exchange information with others. The data showed that over 97% of individuals communicated for refusing, obtaining, and social interaction purposes; 22% would exchange information; and 5% of individuals would comment (this was the function of communication with the lowest prevalence). This is consistent with the past literature.

A 'communication phenotype' in Angelman syndrome has been proposed.[40] Duker et al.[41] compared manding (requesting and rejecting), tacting (describing and labelling), and echoic imitation in individuals with Down syndrome, Angelman syndrome, and pervasive development disorder. For Angelman syndrome, within-syndrome analysis showed strengths in manding, and there were significant differences between manding and echoic imitation. When compared to the other groups, no differences between the levels of manding and tacting were found; however, there were significant differences for echoic imitation, with individuals with Angelman syndrome scoring lower on this domain. However, the Down syndrome and pervasive development disorder groups had some speech, whereas the Angelman syndrome group were completely non-verbal; therefore, these factors may account for some of the differences found for echoic imitation.

As an extension of this research, Didden et al.[40] used the same measure in a larger sample of individuals with Angelman syndrome and a comparison group of individuals with severe or profound intellectual disabilities of a heterogeneous cause. Like Duker et al.,[41] their results showed that manding was the strongest domain, with limited skills in tacting and echoic imitation. When compared to others with severe or profound intellectual disability, manding was of a similar level. However, significant differences were apparent for tacting and echoic imitation, with individuals with Angelman syndrome scoring lower on these domains. No information was provided about any differences in participant demographics of these groups; thus, it cannot be concluded that these findings for Angelman syndrome are unusual or influenced by another factor that was not controlled for.

In addition to descriptive results, factors associated with the functions of communication in Angelman syndrome have also been investigated. The presence of epilepsy, use of anticonvulsant medication, and level of intellectual disability were associated with fewer communicative functions.[40] Moreover, genetic aetiology was related to the use of requesting and commenting, with individuals with non-deletion aetiologies showing a wider variety of communicative functions.[25]

The literature, although limited, provides a detailed description of the profile of communicative functions in Angelman syndrome. Using within-syndrome comparisons, areas of strength and weakness have been identified, which are further understood through comparisons to other genetic syndromes. Since all of the research focusing on communicative functions has used parental reporting, future research should use direct observation strategies to corroborate these findings.

### Communication interventions

There is a paucity of literature exploring the implementation of communication interventions in Angelman syndrome. To date, two intervention studies have been conducted, both focusing on teaching children alternative forms of communication.

The only intervention designed to increase communication skills specifically in Angelman syndrome is ENGs.[42]

ENGs build on the reported preference for the use of gestures in individuals with Angelman syndrome. An ENG develops from a natural gesture or action that is associated with a particular event, which is then shaped to become an intentional, distal gesture that can be understood by a wide range of communication partners (e.g. mimicking pressing a button on a remote control to communicate switching a television channel). Calculator[42] explored the feasibility and acceptability of a home-based intervention programme for nine families who taught their children with Angelman syndrome to use ENGs. Feedback from parents indicated that the programme was both effective and feasible, with children learning to use ENGs as a form of efficient communication. However, weaknesses highlighted in the intervention were the length of time it took for a child to acquire and use ENGs and the time management required, which led to disruption of family life. However, these were not of significant concern to the parents. Further studies showed that when this intervention was implemented in 21 children with Angelman syndrome, all acquired the use of ENGs and exceeded the programme and parents' expectations, with some children also spontaneously using their ENGs.[43,44] These findings indicate that this may be an effective and acceptable intervention for children with Angelman syndrome to increase communication.

Another type of intervention used with children with Angelman syndrome is functional communication training. Radstaake et al.[45] implemented functional communication training for three children with Angelman syndrome to reduce challenging behaviour. Analysis showed that when children were taught a more adaptive form of communication, challenging behaviour decreased for all three children with large-to-medium effect sizes, indicating functional equivalence. The replacement communication methods implemented were speech-generating devices for two of the children and a picture exchange communication system for the third child. This intervention demonstrates that some children with Angelman syndrome can be taught to use AAC and communicate their needs through other forms of communication, alongside highlighting the importance of contingent responses when teaching new forms of communication.

### Methodological limitations

As recommended by Cross and Hare,[29] a quality score in the upper tertile reflects 'reasonable' methodological quality. For the amended criteria used in this scoping review, this indicates a score greater than 8, which was achieved by only two of 13 articles.[40,41] Four papers were in the lower tertile[22,26,27,32] and seven papers were in the mid-tertile.[19,24,25,30,31,33,34]

The most prevalent method adopted in the literature was parental reporting. Ten studies used parental reporting to gather data, two of which used this method in conjunction with direct assessment. Furthermore, most informant questionnaires were not standardized measures. Four studies designed the questionnaire for the project and did not provide any psychometric data, apart from Calculator[19] who appraised the face validity of the questionnaire with AAC experts. A further four studies used measures for which psychometrics had been appraised but not published.

When the psychometrics of a questionnaire have not been appraised, the main shortcoming is that validity and reliability are not ascertained. However, there is a paucity of appropriate measures to explore the topics that these studies are researching. Many measures require some level of verbal ability or are designed for very young, typically developing children; therefore, they may not be appropriate for older children or adults with intellectual disabilities, and so the use of 'unique' questionnaires is understandable. With regard to parental reporting, this relies on parental interpretation of their child's communication, making it more subjective than direct assessment. However, this method allows for the recruitment of a larger sample, which is hard to achieve because of the rarity of Angelman syndrome and is more ecologically valid.

The rarity of Angelman syndrome may mean that there is sample overlap in studies (namely in the Calculator,[19,30,31] Duker et al.,[41] and Didden et al.[40] studies). This may limit the results with regard to generalization. However, because recruitment is problematic and statistical analyses may require large numbers for statistical power, it is advantageous for researchers to get as much data from one sample as possible.

Another issue within the literature is the lack of comparisons to other groups, both typically developing children and children with genetic syndromes and/or intellectual disabilities. The only two studies where comparisons were made were Duker et al.[41] and Didden et al.,[40] although the differences between the groups were not tightly controlled for. Hodapp and Dykens[46] highlighted the importance of having comparison groups in genetic syndrome research since they allow for a greater understanding of what is syndrome-related and what might be accounted for by the level of intellectual disability or other characteristics.

A limitation that has become recognized more recently is the tendency to group individuals with Angelman syndrome regardless of their genetic aetiology. With research highlighting significant differences between individuals with and without deletions, it is important to consider this when results differ from those in the previous literature or when results are unexpected. Earlier research would not have been able to account for this because of the lack of appropriate genetic testing. However, more recent research needs to take this into account when drawing conclusions, since conclusions made at the syndrome level may not be representative of the nuances in communication skills across the different genetic aetiologies of Angelman syndrome, especially given the lower prevalence of non-deletion aetiologies.

## DISCUSSION

The communication profile of Angelman syndrome is characterized by the predominant use of non-symbolic

forms of communication, namely gestures, with strengths in requesting relative to commenting and imitation. Symbolic forms are used by individuals but on a less frequent basis than non-symbolic forms, and the majority of individuals with Angelman syndrome use, or have access to, an AAC device; many are successful in communicating to some degree. Furthermore, it has been shown through intervention studies that children with Angelman syndrome can acquire alternative forms of communication.

The communication profile appears to differ across genetic aetiologies. Individuals with non-deletion aetiologies have better communication skills, particularly in the use of symbolic forms of communication and AAC devices. This is consistent with the wider literature on adaptive behaviour in Angelman syndrome, which shows that individuals with non-deletion aetiologies have higher receptive language scores than those with deletions.[16,18] Therefore, greater use of advanced forms of expressive communication in individuals with non-deletion aetiologies could be reflective of receptive language, even though speech production is similar to individuals with a deletion aetiology. This suggests that communication deficits may just be restricted to speech production in Angelman syndrome and that wider communication skills may be appropriate for the developmental level. However, this possible dissociation has not been directly explored or quantified in the literature, which is a necessary step to substantiate claims of isolated speech production impairment due to the genetic bases of Angelman syndrome.

Addressing the main methodological limitations of the literature, such as the lack of comparison of the communication profile of Angelman syndrome between genetic aetiologies and comparison groups, will also develop a greater understanding of the nuances of the communication profile. Through these comparisons, the specificity of speech impairment can be assessed. Future research should also focus on using direct assessment to investigate the communication profile of Angelman syndrome.

Use of AAC in Angelman syndrome and communicative functions are areas less focused on in the literature. On the other hand, with regard to the functions of communication, the quality of the research is high. Further replication is required to substantiate the claims of a 'communicative phenotype'.[40] Regarding research on AAC, further information about the use of devices and the factors that influence acceptance and success must be gathered. While these areas of research are important, the primary focus should be on detailing the specificities of the communication impairments in Angelman syndrome. Once these are better understood, the impact and downstream consequences for interventions and AAC will be better informed.

Limitations of the present review include the strict inclusion criteria of reviewing studies with the main aim of exploring communication in Angelman syndrome. Thus, other findings about communication profile studies that research the wider clinical profile of Angelman syndrome are absent. However, because of the methodology used, it is assumed that the most important and general findings would have been found and reported in direct studies of communication.

Overall, the findings from the literature indicate a striking and unusual communication profile in Angelman syndrome. When considering the minimal or absent speech of individuals, other forms of expressive communication appear more developed than expected, although more research is needed to quantify this possible dissociation. Emphasis needs to be given to the diversity in profiles among the genetic aetiologies, especially given the homogeneity of the lack of speech, which holds important theoretical and clinical implications.

### Clinical implications

There is debate about focusing on an individual's genetic syndrome with regard to health care approaches and 'labelling'. Indeed, the National Institute for Health and Care Excellence guidelines[47] have increasingly emphasized the importance for a person-centred approach in clinical practice for individuals with intellectual disabilities. However, research has also highlighted the usefulness of recognizing an individual's diagnosis when providing personalized health and social care.[48] For example, several studies have identified the differing profiles of communication between genetic syndromes, which suggests that the genetic basis plays a role in the nature of impairments.[5,6,49] Martin et al.[6] showed differing profiles across a number of language domains for individuals with fragile X and Down syndromes. Furthermore, within syndrome groups certain factors influenced language impairments, for example, the presence of autism in fragile X syndrome for pragmatic language skills. The authors argued that the relative strengths and weaknesses in language of these syndromes were important to consider for assessment and intervention. Therefore, this suggests that by acknowledging a person's diagnosis, an insight can be gained as to the impairments experienced and the possible causes, so that an appropriate and informed decision about support can be made.

Because of the lack of speech that is typical of the syndrome, individuals with Angelman syndrome represent a population that is highly likely to require access to speech and language therapy services. Due to the rarity of the syndrome, the characteristics and communication profile of individuals may not be well known. Emphasis needs to be placed on the homogeneity of the absence of speech across individuals and the apparent discrepancy with other forms of expressive communication; regardless of the variability in the syndrome's characteristics, the vast majority of individuals do not develop spoken language. This strongly suggests a specific expression of the genetic disorder; hence, taking the diagnosis into account is paramount for assessment and intervention which, when combined with individual characteristics, will provide an informed person-centred approach.

## SUPPORTING INFORMATION

The following additional material may be found online:

**Figure S1:** Flow diagram detailing the search process used to select articles for the scoping review.

**Table SI:** Studies exploring communication in Angelman syndrome

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ec Editor's Choice

## Editor's choice

My Editor's Choice for the November 2019 issue is one of the first scoping reviews we have published and has wide implications for clinical research and practice in developmental disability. It focuses on Angelman syndrome, a rare but paradigmatic genetic neurodevelopmental condition originally described by Harry Angelman in *Developmental Medicine & Child Neurology* in 1965. Based on often fragmentary reports of communicative abilities of individuals with Angelman syndrome, the authors manage to usefully delineate modes and functions of communication within the behavioural phenotype, and suggest how knowledge of these can enhance person-centred intervention.

## RESUMEN

### COMUNICACIÓN EN SINDROME DE ANGELMAN: UNA REVISION PANORÁMICA

**OBJETIVO** Se realizó una revisión panorámica para examinar y evaluar datos empíricos sobre el perfil de comunicación del síndrome de Angelman más allá de la disociación descrita entre lenguaje receptivo y habla.
**METODO** Se realizaron búsquedas en tres bases de datos (PsycINFO, Embase y Web of Science) para recuperar artículos que investigan la comunicación en el síndrome de Angelman. Se encontraron 17 artículos que investigan el perfil de comunicación más amplio; Su metodología fue evaluada con criterios de calidad.
**RESULTADOS** A pesar de la ausencia de habla, las personas con síndrome de Angelman tienen un amplio repertorio de conductas comunicativas no verbales, que se caracterizan principalmente por gestos, aunque algunas personas utilizan formas avanzadas como la comunicación simbólica. El uso de formas comunicativas difiere entre las etiologías genéticas del síndrome de Angelman; Los individuos con etiologías sin deleción suelen tener mayores habilidades comunicativas.
**INTERPRETACION** El perfil de comunicación más amplio del síndrome de Angelman se caracteriza por capacidades diversas y multimodales, incluido el uso de formas simbólicas de comunicación que parecen atípicas dada la ausencia de habla. Esto sugiere una probable disociación entre el habla y otras formas expresivas de comunicación, lo que indica un deterioro aislado en la producción del habla. Esto resalta la necesidad en esta población de considerar comunicación alternativa y aportes específicos de servicios diseñados para respaldar los matices del perfil de comunicación del síndrome de Angelman.

## RESUMO

### COMUNICAÇÃO NA SÍNDROME DE ANGELMAN: UMA REVISÃO DE ESCOPO

**OBJETIVO** Uma revisão de escopo foi realizada para examiner e avaliar os dados empíricos sobre o perfil de comunicação na síndrome de Algelman além da descrita dissociação entre liguagem receptiva e fala.
**MÉTODO** Três bases de dados (PsycINFO, Embase, e Web of Science) foram pesquisadas para encontrar artigos que investigassem comunicação na síndrome de Angelman. Dezessete artigos investigando o perfil de comunicação de maneira mais ampla foram encontrados; sua metodologia foi avaliada com relação a critérios de qualidade.
**RESULTADOS** Apesar da ausência de fala, indivíduos com síndroem de Angelman têm amplo repertório de comportamentos comunicativos não-verbais, principalmente caracterizados por gestos, embora formas avançadas como comunicação simbólica sejam usadas por alguns indivíduos. O uso de formas comunicativas difere entre etiologias genéticas da síndrome de Angelman; indivíduos com etiologias de não-deleção tipicamente têm maiores capacidades comunicativas.
**INTERPRETAÇÃO** O perfil de comunicação mais amplo na síndrome de Angelman é caracterizado por capacidades diversas e multimodais, incluindo algum uso de formas simbólicas de comunicação que parecem atípicas dada a ausência de fala. Isso é sugestivo de uma provável dissociação entre fala e outras formas expressivas de comunicação, indicando uma deficiência isolada na produção da fala. Isso enfatiza a necessidade, nesta população, de comunicação alternativa e orientação específica de serviços que dêem apoio às nuances do perfil de comunicacão da síndrome de Angelman.