Communication in Angelman syndrome: A scoping review

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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 seems 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.

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.

Expressive language delay and communication impairments are characteristic of individuals with intellectual disabilities (ID), 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 ID, including vocabulary comprehension, prelinguistic communication and parental response contingency.[1,3,4] These factors can account for variability seen in expressive language development in ID, as well as highlighting key areas that could be targeted for early intervention in order to improve communication abilities.

In ID 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 ID, 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 communication development of infants with Down and Williams syndromes. Findings showed differing communicative 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 and furthermore there was no difference in the total number of words spoken between the groups. This highlights both the theoretical importance of recognising genetic aetiologies associated with communication difficulties in terms of understanding of the roles that gene disorders may play in language and communication and the clinical importance for tailoring specific and effective interventions, possibly at a syndrome level.[6]

One syndrome associated with ID and in which 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 gene UBE3A, and has an estimated prevalence of between 1 in 12,000 and 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 65-75% of individuals. 5-11% of individuals have Angelman syndrome due to a mutation of UBE3A; paternal uniparental disomy (UPD) of chromosome 15, occurs in around 3-7% of individuals and 3-5% of individuals with Angelman syndrome have an imprinting defect.[10,11]

Clinical features of Angelman syndrome are severe to profound ID, 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 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 71-90% of individuals never or rarely producing any speech.[19,20] Of those individuals who do have speech, vocabulary is limited, with literature reporting a spoken vocabulary size of between two and fifteen words, with an average of five.[21–24] However, these words are reported to be rarely used for communicative purposes.[25] Furthermore, it has been suggested that level of ID cannot account for the absence of speech[26,27] because the receptive language abilities of individuals would typically be associated with the presence of expressive language.[27]

Studies investigating phenotypic differences between genotypes in general adaptive skills, including communication, have highlighted that individuals with non-deletion aetiology having significantly greater receptive language skills in comparison to individuals with a deletion. In contrast, the markedly reduced level of expressive language appears homogenous across the genetic aetiologies which suggests that UBE3A may be implicated in speech production[16,18] and further strengthens the argument that the 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 the specificity of impairments would enable a better understanding of possible targets for alternative communication methods as well as effective intervention. Furthermore, due to the genetic basis of this syndrome, understanding the communicative profile of Angelman syndrome has broader implications for understanding of the development of communication and speech production specifically.

The aim of this scoping review is to search published literature in order to summarise 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.

Search Strategy

A systematic search of three databases (EMBASE, PsycInfo and Web of Science) was conducted. The key words 'communicat*' and 'Angelman syndrome' were combined using

the Boolean operator 'AND' to retrieve any papers published between 1960 and October week 4, 2018. No a priori protocol was registered, but further information on the process can be obtained on request to the corresponding author. Figure 1 shows the search process and retrieval of the final papers used in the review.

Study Selection

To be included in the review, papers must have been published in a peer-reviewed journal between 1960 and October 2018. Only published research was considered to ensure that the validity of the findings summarised had been subjected to peer-review. Eligibility criteria required papers to be written in English. One of the main aims of the paper was to explore 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, as this is already widely acknowledged and forms part of the diagnostic criteria.[12] Case studies were excluded due to typical sampling biases from focusing on atypical cases and not providing data at a group level. Two reviewers (EP & RR) independently completed the search process and screening procedure to verify the accuracy of the selection of studies.

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Results

Search results

A total of 17 papers were retrieved which are presented in table 1 in order of the themes discussed within the review. Themes were chosen and agreed upon based on 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, functions of communication, communication interventions. Findings from the studies were charted under the relevant theme and then were further grouped based on common topics for synthesis, for example findings related to gesture use were grouped under the theme of forms of communication.

The methodological quality of papers was assessed based on amended criteria from Cross & Hare[28], which were originally created based on reported best practices for behavioural phenotype methodology. Studies were rated from zero to two on six areas; control group, sample size, recruitment, syndrome diagnosis, methodology and appropriate statistics/comparisons and a total score was obtained for each paper (see table 1). Four papers that implemented communication interventions were excluded from this evaluation as the criteria were not appropriate for intervention designs, and so 13 papers remained for evaluation. Inter-rater reliability was assessed for 46.2% of the included papers (35.3% of total papers), with kappa = .85, thus reliability was excellent.

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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 augmentative and alternative communication (AAC).[19,22,25,29–31] The most commonly used forms reported in the literature come under non-symbolic communication, such as non-speech vocalisations, physical manipulations of others, and gestures[19,22,25–27,31]. These forms of expressive communication are evident regardless of genetic aetiology of Angelman syndrome.[19]

Gesturing is reported to be the most prevalent form of communication in individuals with Angelman syndrome[19,25,27,31] even when the individual is using more advanced forms of communication, such as an AAC device[29,31]. Early research by Jolleff & Ryan[26] stated that only five out of their 11 participants with Angelman syndrome would show gesturing as reported by parents using a standardised questionnaire. Further research that has explored different types of gestures show slightly higher prevalence rates, with estimates for deictic gestures[a] between 50-100%[24,31,32] and specific deictic gestures, such as pointing, being used by between 40-55%[22,31] of individuals with Angelman syndrome and reaching for desired objects used by 75%.[22] Representational gestures, however, do not appear as common as deictic gestures in Angelman syndrome.[24,32]

With regard to the different aetiological causes of Angelman syndrome and the use of gestures, Jolleff et al.[33] reported that individuals that have non-deletion aetiology had 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

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[a] Deictic gestures establish reference by directing attention or indicating an object or event.

study with a larger sample size, there were no significant differences between the genotypes 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 of gestures 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 & Downing[22] found that 50% of parents reported that their child with Angelman syndrome used physical manipulation to communicate. This finding is further corroborated by Calculator[19] in a larger sample of 182 parents citing that around 60% of individuals used physical manipulation 'often' or 'very' often. Similarly, around 60% of individuals used non-speech vocalisations to communicate[19], which have been further detailed as very simple vocalisations that are characteristic of typically-developing children between 0 and 6 months old.[32] Other non-symbolic forms of communication reported in the literature are body movements, facial expressions, and eye gaze.[25,31]

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[19,22,25,31]. Earlier research by Jolleff & 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 use of symbolic communication is greater than previously described. 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 ten signs[22]. The reasons as to 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 individual. As well as signs, Quinn & Rowland[31] also found that other forms of symbolic communication such as concrete symbols, for example using a picture (e.g. picture of a drum to represent music) or an object (e.g. a shoelace to symbolise a shoe) are used by between 18-47% of individuals with Angelman syndrome. Abstract symbols, such as a line drawing of a reaching hand to represent help, were also used by 13-30% of individuals.

There are also significant differences in use of symbolic forms of communication between the genetic aetiologies of Angelman syndrome[19,25] which contrasts with the lack of differences found for non-symbolic forms. Calculator[19] used parental report to explore the differences in forms of communication across individuals with and without deletions. Results highlighted significant differences for the use of enhanced natural gestures (ENGs, discussed later), signing and speech, with individuals with non-deletion aetiologies using these methods more frequently. This is further supported by Didden and colleagues[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 amongst individuals. Didden et al.[25] reported that individuals with severe ID signed more than those with profound ID. In addition, they found that individuals who experienced epilepsy were less likely than those without epilepsy to use symbolic forms of communication. Further to this, 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.[34] However, Didden et al.[25] did not consider level of ID as a

covariate in this analysis. This is an important aspect to consider given that the link between epilepsy severity and level of ID is well established in other genetic syndromes associated with high rates of epilepsy (e.g. tuberous sclerosis complex[35,36]).

In addition to research reporting on the prevalence of forms of communication, Calculator[30] investigated 174 parents' perceptions 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 individuals with Angelman syndrome even when considered against symbolic and AAC forms of communication. In particular, gestures were rated by 84% of parents as very or extremely important, followed by non-speech vocalisations by 76% of parents and physical manipulation by 72%. From this, Calculator[30] highlighted in his conclusion 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 identification of factors explaining variability in the presence and use of different forms. However, the results focus predominantly on occurrence of forms based on informant report. 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 give better understanding of how skills may be transferred and utilised to alternative communication methods.

Use of Augmentative and Alternative Communication

Augmentative and alternative communication (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 & Goldbart[37]).

Aided forms of AAC, both low and high-tech, are reported to be used by between 62-70% of individuals with Angelman syndrome.[19,22] Calculator[19,30] reported a wide range of electronic AAC devices used by individuals with Angelman syndrome, with individuals tending to use more than one device for communicating. With recent advances in mobile technology, there has been a shift towards utilising apps on tablets as a means of individuals communicating, with 48% of individuals with Angelman syndrome using an AAC app on a tablet as their most advanced AAC device.[30] Alongside citing the devices used, Calculator[19] further reported differences in the AAC systems used between individuals with a deletion and those with a non-deletion aetiology. Individuals with deletions were reported to use less advanced electronic AAC systems such as single-message voice output communication aids and were less likely to progress onto 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[30] 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. However, the factors such as support that 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 or not an individual 'accepts' 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.[38] Calculator[19] reported around 60% of individuals with Angelman syndrome accept their most advanced

AAC device and using through thematic analysis of parental report, highlighted a number of factors that influence an individual's acceptance of advanced AAC devices.[29] 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 rejection of the device was the individual's lack of understanding or valuing the device as a communication tool e.g. seeing it as a toy more than a means of communication. Therefore, it was concluded that in order for an individual to accept a device and use it as an effective form of communication the complexity device has to match their developmental level.

Despite advances in mobile technology and AAC devices, non-symbolic forms are still the preferred method of communication by individuals with Angelman syndrome[29,30] as well as symbolic forms which some individuals will choose to use over their AAC device. Furthermore, the majority of 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. However, a smaller proportion of parents viewed AAC devices as very or extremely important compared to the importance of non-symbolic communication.[30]

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. This 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 acceptance and success of devices alongside devising appropriate learning strategies for individuals.

Functions of Communication

Although explored less than the forms of communication in Angelman syndrome, a small number of studies have investigated functions of communication such as the use of protoimperatives (e.g. requesting) and protodeclaratives (e.g. directing another's attention, sharing of experiences). There is a consensus that requesting is the most well developed and most widely used function whereas imitation and commenting appear to be areas of comparative weakness in the communication profile.[25,27,31,39,40] Didden et al.[25] explored the widest range of communicative functions in individuals with Angelman syndrome using parental report. 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, requesting and commenting, the latter providing evidence of use of protodeclaratives in Angelman syndrome. Furthermore, it was found that requesting information and imitation were the least developed areas of communication.

These finding were further corroborated in 300 individuals with Angelman syndrome. Quinn and Rowland[31] used a data mining strategy to explore what 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 and 22% would exchange information. Five percent of individuals would comment, which was the function of communication that had the lowest prevalence, which is consistent with past literature.

A 'communication phenotype' in Angelman syndrome has been proposed.[39] Duker et al.[40] compared manding (requesting and rejecting), tacting (describing and labelling) and echoic imitation in individuals with Down syndrome, Angelman syndrome and pervasive development disorder (PDD). For Angelman syndrome, within-syndrome analysis showed strengths lay 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 but there were significant differences for echoic imitation with individuals with Angelman syndrome scoring lower on this domain. However, the Down syndrome and PDD 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.[39] used the same measure in a larger sample of individuals with Angelman syndrome and a comparison group of individuals with severe or profound ID of a heterogeneous cause. Similarly to Duker et al.[40], 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 those with Angelman syndrome scoring lower on these domains. However, no information was provided about differences between the participant demographics of these groups and so it cannot be concluded that these findings for Angelman syndrome are unusual or are 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 ID were reported to be related to fewer communicative functions.[39] Moreover, the genetic aetiology was related to the use of requesting and commenting with those with non-deletion aetiologies showing a wider variety of communicative function.[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 further understood through

comparisons to other genetic syndromes.   Future research should focus on intervention strategies to encourage the development of weaker communicative functions and increasing the variety of functions an individual communicates for.

Communication Interventions

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

The only intervention designed to increase communication skills specifically in Angelman syndrome is Enhanced Natural Gestures (ENGs).[41]   ENGs build upon the reported preference for use of gestures among 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 variety communication partners (e.g. mimicking pressing a button on a remote control to communicate switching the television channel). Calculator[41] explored the feasibility and acceptability of a home-based intervention programme for nine families that taught their children with AS 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 child to acquire and use ENGs and the time management required which led to disruption of family life but these were not reported to be of significant concern to the parents. Further studies showed that when this intervention was implemented with a total of 21 children with Angelman syndrome, all acquired the use of ENGS and exceeded programme and parents' expectations with some also spontaneously using their ENGs.[42,43]

These findings indicate that this may be an effective, 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.[44] implemented Functional Communication Training for three children with Angelman syndrome in order to reduce challenging behaviour. Analysis showed that when the 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 system for the third child. This intervention demonstrates that some children with Angelman syndrome can be taught to use AAC and to 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 & Hare[28], a quality score in the upper tertile reflects 'reasonable' methodological quality. For the amended criteria used in this review, this indicates a score greater than eight, which was only achieved by two of 13 papers.[39,40] Four papers were in the lower tertile,[22,26,27,31] and the majority of seven papers were in the mid-tertile.[19,24,25,29,30,32,33]

The most prevalent method utilised in the literature was informant report. Ten studies used informant report to gather data, two of which used this method in conjunction with direct assessment. Furthermore, the majority of the informant questionnaires were not standardised 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 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 and therefore, may not be appropriate for older children or adults with ID, and so the use of 'unique' questionnaires is understandable. With regard to parental report this relies on parents' interpretation of their child's communication making it more subjective than direct assessment. However, this method allows for recruitment of a larger sample which is hard to achieve due to the rarity of Angelman syndrome and is more ecologically valid.

The rarity of Angelman syndrome may mean there is sample overlap in studies (namely in Calculator[19,29,30] and the Duker et al.[40] and Didden et al.[39] studies). This may limit results with regard to generalisation. 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 and other groups with genetic syndromes and/or intellectual disability. The only two studies in which comparisons were made were Duker et al.[40] and Didden et al.[39], although differences between the groups were not tightly controlled for. Hodapp and Dykens[45] highlight the importance of having comparison groups in genetic syndrome research as they allow for a greater understanding of what is syndrome related and what might be accounted for by level of intellectual disability or other characteristics.

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

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 the communication profile studies researching the wider clinical profile of Angelman syndrome are absent. However, it is assumed that the most important and general findings would have been found and reported in direct studies of communication due to the methodology used.

Summary

In summary, the communicative profile of Angelman syndrome is characterised 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, with many being 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 communicative profile appears to differ across the genetic aetiologies, with individuals with non-deletion aetiologies being more able in their communication abilities, particularly in the use of symbolic forms of communication and AAC devices. This is consistent with the wider literature describing 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. This highlights that communication deficits may just be restricted to speech production in Angelman syndrome and wider communication skills may be appropriate for 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 an isolated speech production impairment which is due to the genetic basis of Angelman syndrome.

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

Use of AAC in Angelman syndrome and communicative functions are areas less focused on in the literature. However, for functions of communication, the quality of the research was high but further replication is required to substantiate the claims of a 'communicative phenotype'.[39] With regard to research on AAC, further information about the use of devices and factors influencing acceptance and success needs to be gathered. Whilst these areas of research are important, the primary focus should be on detailing the

specificities of the communication impairments in Angelman syndrome, as once these are better understood, the impact and downstream consequences for interventions and AAC will be better informed.

Overall, findings from the literature indicate a striking and unusual communicative profile in Angelman syndrome, that 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 the profile amongst the genetic aetiologies, especially given the homogeneity of the lack of expressive speech, which holds both important theoretical and clinical implications.

Clinical implications

There is debate about focusing on an individual's genetic syndrome with regard to healthcare approaches and 'labelling'. Indeed, National Institute for Health and Care Excellence (NICE) guidelines[46] have increasingly emphasised the importance for a person-centred approach in clinical practice for individuals with ID. However, research has also highlighted the utility of recognising an individual's diagnosis for the provision of personalised health and social care.[47] For example, other research has identified differing profiles of communication between genetic syndromes that suggests the genetic basis plays a role in the nature of impairments.[5,6,48] Martin et al.[6] showed differing profiles across a number of language domains for individuals with fragile X and Down syndromes. Furthermore, it was highlighted that 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 production of these syndromes were important to consider for assessment and

intervention. Therefore, it 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.

Angelman syndrome is a population that is highly likely to require access to speech and language therapy services because of the lack of speech in the syndrome and, due to the rarity, the characteristics and communicative 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 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 and 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.

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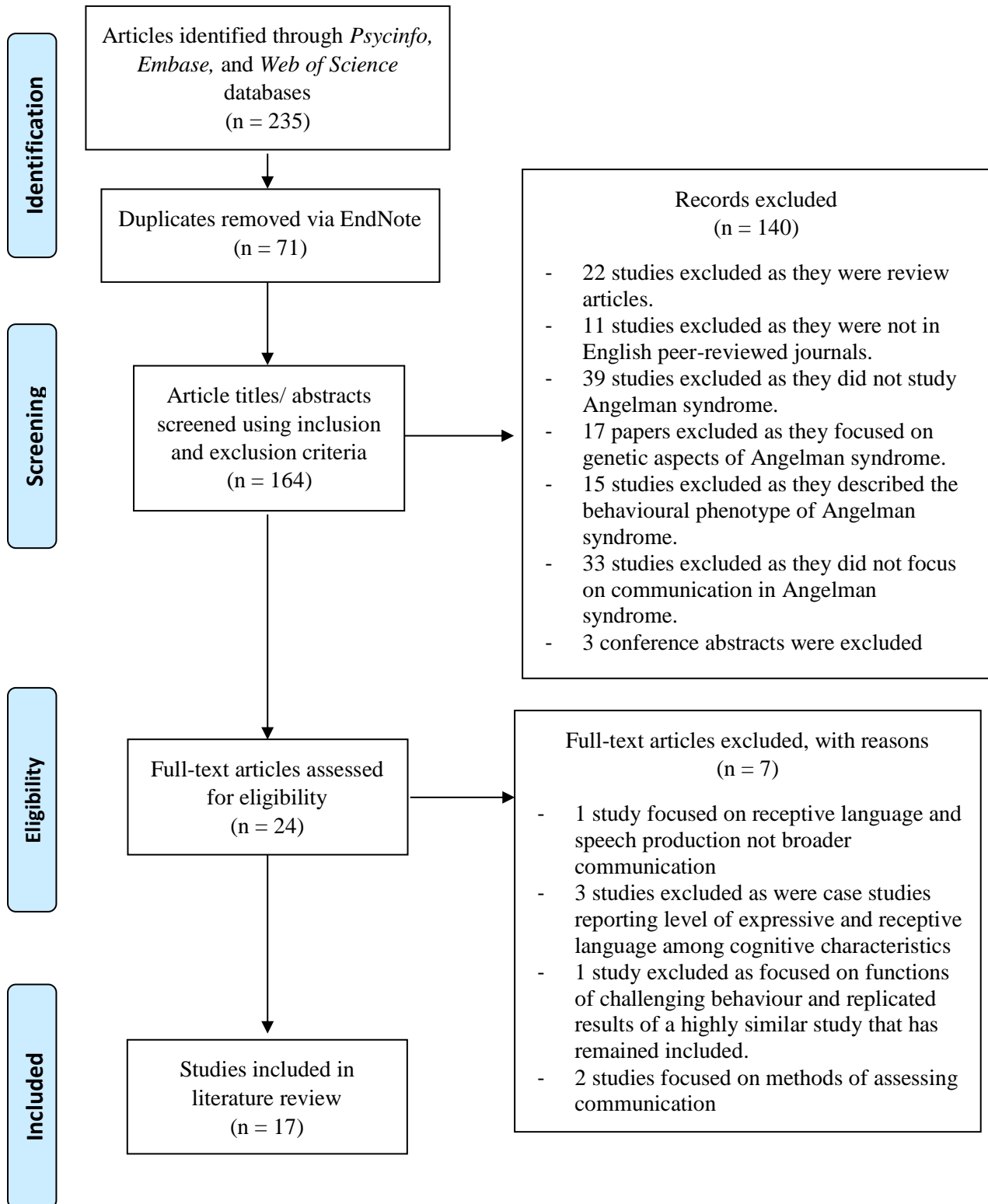
Figure 1.
Flow diagram detailing the search process for selecting papers for the scoping review.

Table SI. *Studies exploring communication in Angelman syndrome*

Citation	*N*	Age range (mean)	Level of ID	Genetic mechanism (%)	Design	Measures used	Outcome variables	Quality score (out of 12)
Forms of communication								
Alvares & Downing[22]	20	1.5-13 yrs (6.1)	Not specified	55% deletion, 35% no visible deletion, 10% UPD	Online parent survey	Informal, non-standardised questionnaire	Forms of communication	3
Micheletti et al.[24]	10	5-11 yrs (7.1)	Severe	70% deletion, 30% UBE3A mutation	Parent survey	Italian version of MacArthur-Bates Communicative Development Inventory (CDI)[b]	Level of comprehension and speech production Forms of communication	5
Jolleff & Ryan[26]	11	2.5-15.3 yrs[a]	Not specified	Not specified	Pilot study - Direct assessment	Receptive-Expressive Emergent Language (REEL) test Modified version of Preverbal Communication Schedule (PVCS)	Level of receptive and expressive language Forms of communication	3
Penner et al.[27]	7	12-40 yrs (28.3)	Profound	Not specified	Direct assessment	Prelanguage Communication Assessment for Cognitive, Social and Expressive Generic Skills (PCA)	Level of sensorimotor cognitive development and expressive language Forms of communication Oral-motor findings	2
Grieco et al.[33]	9	2.8-10.5	Not	88.9% deletion,	Direct	Vocalisation analysis	Vocalisation production	6

		yrs (7.8)	specified	11.1% UBE3A mutation	assessment	Gesture coding	Patterns of gesture use	
Jolleff et al.[34]	14	7-21 yrs[a]	Not specified	50% deletion, 35.7% UPD, 14.3% imprinting (grouped as non-deletion)	Parent interview and direct assessment	CDI The pragmatics profile Cass and Lees assessment schedule Paediatrics oral skills package	Gentoype x phenotype differences in communication	6

Augmentative and Alternative Communication (AAC)

Calculator[19]	182	<3->18 yrs[a]	Not specified	59% deletion, 41% non-deletion (not further specified)	Online parent survey	Non-standardised questionnaire	Genotype x phenotype differences in methods of communication and AAC use	6
Calculator[30]	122	<3->18 yrs[a]	Not specified	60% deletion, 40% non-deletion (not further specified)	Online parent survey	Non-standardised questionnaire	Differences in methods of communication between those who accepted AAC devices and those who rejected Qualitative reasons for acceptance or rejection of AAC device	5
Calculator[31]	209	<3->18 yrs[a]	Not specified	56% deletion, 12% UPD, 12% mutation, 10% imprinting, 1% other abnormality	Online parent survey	Non-standardised questionnaire	Methods of communication and perceived importance Types of AAC used Use of AAC devices among children	5

Functions of communication

Didden et al.[25]	79	3-66 yrs (19.6)	Profound to moderate	63% deletion, 16% UPD, 1% imprinting, 7% mutation	Parental report	Inventory of Potential Communicative Acts (IPCA)	Methods and functions of communication Correlates of communication impairment	7
Quinn & Rowland[32]	300	0-22 yrs[a]	None to severe	No information available	Professional report	The Communication Matrix	Forms of communication Functions of communication	4
Didden et al.[40]	108 AS, 117 comparison group	2-44 yrs (15.2)	Severe to moderate	15% clinical diagnosis 85% genetic confirmation (78% deletion, 11% UPD, 3% translocation, 8% unknown)	Parental report	VerBAS	Communicative functions used, Within- and between-syndrome data on tacting, manding, and echoic imitation Correlates of communicative functioning	9
Duker, van Driel & van de Bercken[41]	77 (26 AS, 26 DS, 25 PDD)	3.2-52.2 yrs (15.8)	Severe to moderate	Not specified	Parental report	VerBAS	Within- and between-syndrome data on tacting, manding, and echoic imitation	10

Communication interventions

Calculator[42]	9	3-10 yrs (7.1)	Severe to profound	100% deletion	Feasibility of intervention	Non-standardised questionnaire	Strengths and weaknesses of the ENG intervention	N/A
Calculator[43]	18	<3-18 yrs	Not specified	78% deletion, 6% UPD, 1% ICD, 11% unknown cause	Intervention – 'B' design	Non-standardised questionnaire	Goal attainment levels Efficacy of intervention	N/A

Calculator & Diaz-Caneja Sela[44]	3	9, 9 & 10 yrs	Not specified	100% deletion	Intervention – 'B' design	N/A	Illustration of intervention training procedure Efficacy of intervention	N/A
Radstaake et al[45]	3	6, 7 &15 yrs	Severe	33% deletion, 33% imprinting, 33% mutation	Intervention	Functional analysis Functional communication training	Effect sizes of intervention on challenging behaviour Functional equivalence	N/A

[a] No mean age provided by the authors

[b] Only measures in the study that are related to communication are reported.