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Communication development in Angelman's syndrome

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Abstract

The communication development in 11 children with Angelman's syndrome is described. The clinical observation that these children appear to have a greater ability with receptive rather than expressive language is investigated and these skills assessed using published communication schedules. In addition the understanding and the use of non-verbal communication such as natural gesture was studied. The data collected highlight the fact that these children have developed very few words and have difficulty in using gestural or sign systems. This has implications for speech and language therapists and the children's remedial programmes. Possible future longitudinal studies are suggested.

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Angelman's syndrome (or happy puppet syndrome) was first recognised in 1965.[1] Features on electroencephalography have been found to lend support to the diagnosis,[2] leading to increasing recognition of the syndrome.[3] Clayton-Smith reports that 60% of these children have a *de novo* deletion of chromosome 15q11–13 that is visible cytogenetically and inherited from the mother (sporadic with low recurrence).[4] A further 15% with this deletion can only be detected by DNA analysis, a small percentage have a chromosomal rearrangement in the mother involving chromosome 15, and the remainder, around 15%, have no visible deletion (familial with high recurrence).

Collected data from 151 cases reported over the last 24 years have indicated a distinct clinical syndrome.[5] All children with Angelman's syndrome have severe learning difficulties but several authors have commented on the marked lack of expressive speech.[3 6 7] Elian remarks that 'lack of speech was disproportionate to degree of retardation'.[8] The severity and nature of this does not seem to have been detailed but in the 36 children described by Robb *et al*, aged between 18 months and 13·5 years, only seven had any recognisable words and the maximum number of words used was three.[3] In the study of five children by Fisher *et al*, the oldest of whom was 10·7 years, none had developed any recognisable words.[6] In addition there is anecdotal evidence of little or no aptitude for learning a gestural system such as Makaton.

There has been increasing interest in relating genotypes to behavioural phenotypes. This paper briefly reviews the clinical features and describes a pilot study, undertaken to look more specifically at the language difficulties of children with Angelman's syndrome. This has implications for treatment and remediation.

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Clinical features

Clinical features include delayed developmental milestones from early infancy, ataxic movements, typical facial features with frequent tongue thrusting, unprovoked laughter, and seizures. Angelman remarked on the resemblance of these children, with their jerky arm movements, to a marionette and used the term 'puppet children' to describe them.[1]

OROFACIAL STRUCTURE

Several facial abnormalities have been identified as common. These include wide mouth, tongue protrusion, widely spaced or irregularly spaced teeth, and a pointed chin. A number of children in the series of Robb *et al* were reported to have had poor chewing or sucking in early infancy.[3]

SEIZURES

In the group studied by Robb *et al* the mean age of onset of seizures was 2 years.[3] Ninety three per cent had developed seizures by 4 years.

MOVEMENT DISORDER

This is present in all children with Angelman's syndrome: ataxia affecting all limbs with a wide based gait and frequent flapping of the hands. In the study of Robb *et al* 69% were walking between 2–3 years.[3]

Pilot study

We conducted a pilot study to investigate language development in 11 children aged 2·5 years to 15·3 years. These children were referred by the neurology team at the Hospital for Sick Children, London.

AIMS OF THE STUDY

(A) Short term

- To describe receptive and expressive language skills.
- To assess the understanding of non-verbal communication.
- To assess the use of non-verbal communication.

(B) Long term

- To document the natural history of language development in these children.
- To consider implications for intervention and treatment.

Methods

We anticipated low functioning children who

would not necessarily comply with formal testing. Therefore we used the following: the Bzoch-League receptive expressive emergent language scales (REEL)[9] and a modified version of the preverbal communication schedule of Kiernan and Reid (PVCS).[10]

The REEL measures language skills, both receptive and expressive, and is banded into age equivalents. The PVCS has both interview and test items, and is aimed at drawing up a profile to aid programme development.

Only eight of the 25 sections of PVCS were chosen. This was to limit the length of the test period and to concentrate on specific aspects of communication. Our particular interest was to look at non-verbal communication, especially through gesture and the ability of these children to imitate motor patterns.

The children were assessed by the authors in a clinical setting mostly by informal observation and consultation with parents.

Results

REEL

Comprehension

Although the children tested were aged 2·5 years to 15·3 years, the highest level of comprehension of language was at an age equivalent of 22 months (table 1). It would appear in our pilot group that the children with Angelman's syndrome were unable to understand more than two key words in a sentence. It should be noted, however, that as the children get older they seem to acquire a larger receptive vocabulary, that is a greater repertoire of single word understanding.

Table 1 Scores on the Bzoch-League REEL scale

Child No	Chronological age (years)	Comprehension (months)	Expression (months)
1	15·3	22	12
2*	13·1	22	12
3	11·5	22	12
4	9·3	18	14
5*	7·8	16	12
6	6·3	11	11
7	6·2	18	11
8	5·8	9	7
9	3·5	22	11
10	2·5	11	6
11	2·8	11	6

*Children 1–3 and 4 and 5 are siblings.

Table 2 Ratings on the PVCS

	Sections†							
Child No	1	4	5	12	17	18	19	24
1	U	U	R	U	R	R	R	U
2*	U	U	U	U	R	U	R	U
3	U	U	U	U	R	R	R	R
4	U	U	U	U	R	U	N	U
5*	U	U	U	U	R	U	N	U
6	U	U	U	N	N	U	R	N
7	U	U	R	R	N	U	N	R
8	U	U	U	N	N	R	N	R
9	U	U	U	N	N	R	N	R
10	U	U	R	N	N	R	N	R
11	U	R	N	N	N	R	N	R

*Children 1–3 and 4 and 5 are siblings.
U=usually, R=rarely, N=never.
†1=Needs and preferences; 4=control of hands and arms; 5=social interaction without communication; 12=motor imitation; 17=communication through gestures; 18=communication through manipulation; 19=communication through pointing; 24=understanding of non-verbal communication.

Expressive speech

There was a marked gap between the expressive and comprehension abilities of some children, especially those who were older (table 1). The children had a notable lack of intelligible speech and, in fact, only the two groups of siblings in our study gained a few words. Many children had not developed any speech. However, the maximum gap between comprehension and expression was only 11 months.

PVCS

In this study we looked particularly at eight sections of the schedule. The first three are all in 'pre-communicative behaviours'. The others are in 'imitative skills', 'informal communicative behaviours', and 'formal communication skills'.

Most of the children had ability in the following areas: expression of needs or preferences and control of hands and arms; they also showed evidence of social interaction without communication (table 2).

Understanding non-verbal communication

Most children had some ability in this area, for example taking an object when it is offered or looking to where someone is pointing. Despite having the ability to understand this form of communication they were unable to use it. This is described below.

Imitation of motor patterns

Only the children with familial Angelman's syndrome (that is the first five listed in table 1) were able to copy clapping, waving, etc and one other child made rare attempts. This may be a significant aspect when considering their use of gesture and other non-verbal sign systems.

Use of non-verbal communication

There are three sections which relate to use of non-verbal communication.

(i) Communication through gesture – This aspect was not fully established even for those children with some Makaton training. Five children used gesture rarely and these were the familial cases. The other six children made no use of gesture.

(ii) Communication through pointing – Four children made partial use of pointing and seven did not point at all.

(iii) Communication through manipulation – Almost all the children made some use of this method of communication; their preferred way of imparting their needs was to take an adult by the hand and guide them to what was wanted. They usually used touch to get attention and pushed a hand away if they did not want help. There was frequently a lack of referential eye gaze and they conduct their communication through very physical channels.

Discussion

It would therefore appear that despite achieving a level of verbal comprehension that normally would be sufficient for developmentally delayed

children to obtain some recognisable speech, those with Angelman's syndrome rarely did so. The older children had acquired a greater understanding of single words and had matured in their social understanding and so appeared more 'worldly wise'; in fact, however, the measurable gap between their comprehension and expressive speech remained small. Despite Clayton-Smith's assertion that children with Angelman's syndrome must communicate by sign language or primitive gesture,[4] we found that they do not appear to be able to use natural gesture or Makaton signs in a functional way to indicate their needs. We noted that the children with some words or Makaton signs tended to be familial cases, and even then the Makaton signs were largely used for labelling or after a direct request to use them. They were not used for spontaneous communication.

The pilot study has raised three important questions: (i) whether these children have difficulties in planning and executing motor activities including speech; (ii) whether there is a more fundamental problem of using communication for social interaction; or (iii) whether the primary difficulty was one of low developmental age.

IMPLICATIONS

There are significant implications for speech therapy practice even at this early stage of investigation. Firstly, children with Angelman's syndrome tend to understand non-verbal communication, so the use of Makaton signing remains potentially useful to aid their comprehension, but attempts to train the use of signing to augment their speech may be unsuccessful due to their poor imitation skills and possible motor organisational difficulties. In one case the introduction of choices on a picture/symbol communication chart has proved more useful than signs. They need to be encouraged to use natural gesture and improve their turn taking and joint attention skills. The children rely on physical manipulation rather than on more subtle forms of communication such as eye gaze to communicate.

We plan to undertake a larger study to look in greater depth at the language and speech development of these children and to compare the familial and non-familial cases. We will need to relate language comprehension level to non-verbal cognitive ability and to assess the possible problems of gross, fine, and oral motor control.

It is a priority to try and establish a level of cognitive function in these children. In particular, an investigation of their visual perceptual skills should help to establish which augmentative system of communication would be most appropriate to their needs. Finally the relationship between levels of comprehension and the use of social functions of communication should be investigated.

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