# 9
# COMMUNICATION

*Bernard Dan*

Speech impairment is one of the consistent features of Angelman syndrome (Williams *et al.* 2006). It has been characterised as 'absence or minimal use of words'. This applies to the great majority of patients. In their survey of 61 patients, Lossie *et al.* (2001) found that no patients with a 15q11–q13 deletion had more than three words. 71% of these patients had no words at all. Patients with a *UBE3A* mutation were similar to them with regard to speech disability. Just over half of the patients with either uniparental disomy or imprinting defect had at least three words. In rare cases, patients may use several dozen words, particularly where the underlying abnormality is uniparental disomy or an imprinting defect (Dan *et al.* 2000a) (see Chapter 4). There is also a category of patients with a mosaic form of an imprinting defect, and these individuals also tend to have better speech abilities, in the context of a possibly variant phenotype (see Chapter 6). Some of them have some words and jargon (non-words) with good prosody (Bonati *et al.* 2007). Some patients with mosaic imprinting defect have many words, and a few patients can put together simple sentences (Gillessen-Kaesbach *et al.* 1999, Brockmann *et al.* 2002, Nazlican *et al.* 2004). Variation in communication abilities may also occur as a result of motivational and experiential factors. Some authors have suggested that lower abilities may correlate with the severity of epilepsy (Didden *et al.* 2004a, Jolleff *et al.* 2006). It must be emphasised that even patients who can say 'many' words virtually never use speech as their primary means of communication.

The severe verbal impairment that is characteristic of Angelman syndrome poses particular problems with regard to communication. It affects socialisation through communicative and other social activities. It has functional links with other typical features, including cognitive impairment (see Chapter 8), motor disorders (see Chapter 10), behavioural features (see Chapter 7), attention deficit (see Chapters 7 and 8), and features involving other oral functions (protruding tongue, tongue thrusting, sucking and swallowing disorders, frequent drooling, feeding problems, abnormal food-related behaviours and excessive mouthing and chewing behaviours). With regard to the role of language in thought, it is likely that speech impairment also contributes to the intellectual disability. It has been suggested that expressive language may be a prerequisite for the emergence of higher-order thought processes, so that severely impaired development of speech would eventually result in intellectual deficits. Reciprocal relationships between speech impairment and intellectual disability seem to be the rule. In particular, speech impairment is a consistent feature in the context of nonspecific severe-to-profound cognitive deficits. Impaired conceptual development may reduce the drive to express meaning. This would mostly affect the semantic



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component of language. However, the spontaneous interest for more or less refined non-verbal language, and the relative preservation of receptive verbal language, suggest that speech impairment is a discrete feature in Angelman syndrome (Penner *et al.* 1993).

In contrast to other features of Angelman syndrome, including some other areas of cognition in which research on animal models (see Chapter 15) could be expected to yield valuable insights, the study of language deficit poses specific problems, which can only be addressed by studying the patients directly, using approaches designed to decipher the complexity of human language. Furthermore, application of the classic modular approach of language with respect to subsystems, such as syntax and semantics, would be very limited and potentially misleading. Even at a superficial level, comparison of phonological, lexical and semantic aspects of the highly restricted language of individuals with Angelman syndrome with those emerging in typical development is unsatisfactory.

**General context of communication**

Almost all patients show openness towards social interaction (see Chapter 7). Patients commonly tend to initiate communication. However, they often show significant deficits in several pragmatic aspects that are important for promoting and maintaining communication. Stability of gaze contact is often markedly perturbed (see Chapter 8). Reduced eye contact may be in relation to hyperactive behaviour (see Chapter 7), but it also occurs in older individuals, who show less or no hyperactivity. Orientation to the speaker may be impaired because of deficit in orienting attention (see Chapter 8). This is partly compensated for by relatively good situational understanding based on perception of contextual cues (despite common errors in attributing meaning to signals). However, joint attention and communicative sharing are commonly perturbed. Reciprocal exchange may be difficult in a rhythmic, dialogue-like fashion. Turn-taking is impaired by impulsivity (see Chapter 7). Communication partners may also show difficulties in developing or maintaining an interaction context propitious for communication, as they do not receive expected signals. Facial expression, body gestures and eventual vocalisation patterns may not express clear communicative intent. In studies of communicative behaviour, Duker *et al.* (2002) and Didden *et al.* (2004a) found that individuals with Angelman syndrome used their communicative function primarily for requesting wanted objects or activities or reject unwanted ones (i.e. manding function) (see below). In contrast, (non-verbal) labelling of objects or activities (i.e. tacting) was much less developed and echoing was virtually absent.

**Receptive verbal language**

Receptive verbal language refers to comprehension of spoken language. Receptive speech and expressive speech involve different processes and neural structures. Receptive verbal skills are often reported to be better than expressive verbal ones (Clayton-Smith 1993, Trillingsgaard and Østergaard 2004, Williams *et al.* 2006), though this notion has been disputed in one study of 20 children aged 2–14 years (Andersen *et al.* 2001). In fact, having a much larger receptive vocabulary than an expressive one seems to be a general rule in human language, as normal individuals commonly recognise many words that they never actually use themselves. Moreover, the level of verbal understanding may be difficult to

appreciate in ('real life') ecological situations, when non-verbal contextual information complements purely verbal cues.

Understanding speech implies prior learning of the meanings of words. This process may rely on two different mechanisms. One mechanism depends on understanding of the intentions of speakers (Bloom 2000). Understanding other people's covert intentions, i.e. 'theory of mind' or metacognition, is typically impaired in autism. Unlike children with autism (see Chapter 6), those with Angelman syndrome do not generally have a primary deficit in theory of mind and do not have a social apprehension. Therefore they can make use of information arising from the social situation such as gaze cues to learn the meaning of words. Another mechanism for learning what words mean involves logical reasoning strategies in order to infer the referents of words (Halberda 2003). One example of reasoning strategy which children with Angelman syndrome tend to use (within the limits of their cognitive abilities) is known as 'mutual exclusivity', i.e. presuming that each object has only one label.

Studies of processing of acoustic language are under way. Our preliminary neurophysiological findings in event-related potentials following presentation of auditory non-language and language stimuli suggested that early encoding of speech sounds showed no marked differences between children with Angelman syndrome and typically developing children in distinguishing known words from non-words (see Chapter 8).

Administering the receptive language scale (scale 4 from the Mullen Scales of Early Learning, see Mullen 1995) in a group of 16 children aged 5–15 years with a 15q11–q13 deletion, Trillingsgaard and Østergaard (2004) found a range of performance corresponding to what is expected at 5–15 months (9.3 ± 2.9 months). Accordingly, in our experience, most patients understand many single words. These include names of familiar objects and of familiar people, as well as simple action words. Words referring to concrete objects are usually more easily understood than those describing concepts or relationships, which may remain unintelligible. In contrast to single words, comprehension of two-word utterances (which is achieved by about 2 years of age in typically developing children) may be limited by poor attention (see Chapter 8) and difficulties in processing. Understanding of small phrases, which typically implicate linguistic concepts, such as size, prepositions and adjectives, may be difficult in relation to intellectual disability rather than specific receptive language impairment.

**Expressive verbal language**

As mentioned above, expressive verbal communication is generally considered to be less developed than receptive verbal communication, except in one study in which both were equally severely impaired (Andersen *et al.* 2001). The discrepancy between receptive and expressive communication may lead to frustration among patients, and this may further decrease the quality of communication.

The first vocal sounds produced by infants relate to crying behaviour. The sounds correspond to intense expiration modulated by tensed vocal cords. Babies with Angelman syndrome may have a high-pitched, cat-like cry (Clayton-Smith 1993). The quality of crying gradually becomes diversified, enhancing the communicative value of crying. However,



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infants with Angelman syndrome tend to cry less than typically developing infants, and they tend to underuse crying in communication contexts. From a developmental perspective, they also show a marked delay and lack of variability in early vocalisation such as cooing and babbling (see Chapter 2). The quality of produced sounds depends on postural behaviour and development. The first non-crying vocalisations appear in association with the emergence of head control. These sounds, which are produced in the supine position, tend to be guttural (e.g. 'rr'). Limited investment of ventral positions and markedly delayed verticalisation seem to be accompanied by limited production of labial sounds (e.g. 'bb', 'mm'). Overall, oral sounds are rare. They may be mostly confined to shrieks and screams, which are sometimes uttered in a context of communication. Some patients develop specific meaningful intonations whose significance can be understood by people who know them well. However, vocalising tends to become less varied and less modulated as the child grows, and many patients lose vocalisation over time. As mentioned above, a significant proportion of patients do not use any words at all (Jolleff and Ryan 1993, Penner *et al.* 1993, Alvares and Downing 1998, Andersen *et al.* 2001, Lossie *et al.* 2001). Others can say a small number of words (usually fewer than five), which tend not to be particularly common ones.

The cause of the virtual absence of speech is yet to be elucidated. The presence of sound excludes aphonia or other laryngeal problems. The developmental aspect rules out aphasia, though a form of developmental dysphasia is likely. Penner *et al.* (1993) suggested that the speech deficit could be related to oromotor dyspraxia. Oral dysfunction (see Chapter 10) has hardly been studied in Angelman syndrome. In a preliminary study of four children with a maternal chromosome 15q11–q13 deletion, Dumont *et al.* (2000) noted that the most consistent features were parted lips, mandibular prognathism, sialorrhoea, low tongue position, tongue protrusion and tongue hypokinesia. They noted improvement of these features when body position was high (vertical, antigravity posture) body position as compared to low (horizontal) body position. These changes mainly concerned tonic control and reduced inertia. The features were generally qualitatively better during voluntary movements than during automatic ones. However, functional coordination of (non-vocal) orofacial movements often appears to be easier for spontaneous movements than on imitation. Common methods for assessing orofacial praxis (Vargha-Khadem *et al.* 1995) cannot be easily applied in patients with Angelman syndrome because of limited cooperation. It seems likely that these observations reflect a contribution of oral dyspraxia, but this requires further documentation. Other features of dyspraxia may be observed in Angelman syndrome (see Chapter 10).

The observation that eventual words are often pronounced unclearly, with sounds limited to vowels or a few consonants, may suggest a contribution of severe dysarthria or anarthria. This could be ascribed to deficits in various motor systems, including cerebellar dysfunction.

**Expressive non-verbal language**

In a study of seven individuals with Angelman syndrome who lived in a residential home, Penner *et al.* (1993) found that expressive communication served primarily to indicate requests as opposed to conveying protest or comments. Similar results were found in a study

of 109 individuals with Angelman syndrome aged between 2 and 44 years (mean 15.2), 67% of whom lived at home with their parents and 33% in a residential facility (Didden *et al.* 2004a). They found that the manding function (i.e. request or reject objects or activities) was relatively well developed, and was at a level similar to that of the control group with nonspecific intellectual disability. However, the overall level of expressive communication functioning was lower in the study group than in the control group. Patients with Angelman syndrome used manding significantly more than tacting (i.e. labelling objects or activities), and tacting more than imitation. Duker *et al.* (2002) found similar results in a group of 26 individuals with Angelman syndrome aged between 3 and 52 years (mean 15.8). They found no statistical differences in the use of mand or tact between this group (26 individuals with Down syndrome aged between 4 and 40 years, mean 11.7) and a group of 25 individuals with pervasive developmental disorder aged between 5 and 38 years (mean 15.1).

None of the seven severely affected patients studied by Penner *et al.* (1993) in a residential institution, and a minority of the eleven children studied by Jolleff and Ryan (1993), used gestures for communication. In a larger survey of 340 individuals with Angelman syndrome (93% of whom lived at home), only a minority (47%) were reported to have expressive abilities (speech or signs) (Walz and Baranek 2006). In contrast, in other experiences including ours, the vast majority of patients develop some degree of visuo–manual expressive communication (Clayton-Smith 1993, Miller 1995, Alvares and Downing 1998, Duker *et al.* 2002). Expressive skills are often multimodal. They typically include facial expression, although an inconsistent bias towards positive expressions is possible, perhaps limiting expressive selectivity. The question of whether laughing is appropriate and serves as a mode of communication is still under debate (see Chapter 7). In our experience, laughter seems to be clearly related to (not always consensually pleasant) contextual precipitants most of the time. At times, however, it may signal anxiety (Bax 2001). Patients often implicate communication partners into their activities by taking them by the hand, though they do not usually 'use' the partner's hand to perform desired tasks as individuals with autism tend to do. Many patients spontaneously develop the ability to point towards objects and to make use of this in communication. Pointing may involve the entire hand, and may lack accuracy, but it often evolves to single-finger pointing. Many patients can nod to signify 'yes', shake their head to signify 'no', or wave their hand with consistency. Other communicative gestures can emerge, either without or with formal learning (Fig. 9.1). Gestures are usually used singly as opposed to coordinated sequences. Learning of communicative gestures may be impaired by attention deficit (see Chapter 8) and motor difficulties (see Chapter 10). Dyspraxia, in particular, may manifest as deficits in the performance of representational gestures, imitation of gestures and gesture sequences. Simplified communicative sign systems taking these aspects into account have been used, raising the important question of the compromise that needs to be achieved to provide appropriately shared sets of codes which would facilitate communication between individuals.

The possibilities of abstraction enable the use of symbols or more figurative drawings as tools in communication in some patients. Individually tailored sets of pictograms can be used for expressive language (Fig. 9.2). Patients can learn to present or choose between




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**Fig. 9.1.** Sign language: (A) 'quiet', (B) 'listen', (C) 'sweetie' ('candy'), and (D) 'apple'.

communication cards or to point to pictures on picture charts or activate electroni
communication devices. Five of the 20 children (aged 1.5–14 years) studied by Alvare
and Downing (1998) used such pictograms, and three used a voice-output system. A survey
of 179 families revealed that while most children used signs and gestures, only 40% use

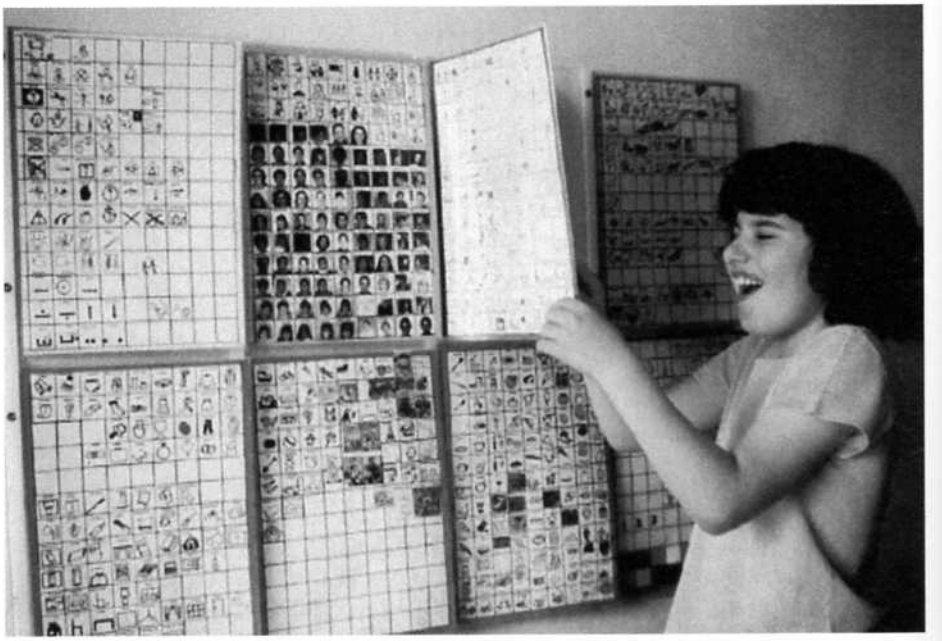

**Fig. 9.2.** Communication device.

picture boards and only 20% used electronic systems (Miller 1995). Adaptation between the individual's needs, abilities, environmental factors and future prospects should be central in designing an approach for optimising communication. In our experience, a number of children who have a sophisticated communication device do not use it to communicate functionally. In addition, a few children learned a sign language that proved to be 'too private' to use after they left their first school and joined another institution where another sign language was in use.

## Conclusion

Communicative functioning is reduced because of intellectual disability, specific dysphasic problems (which may involve oral dyspraxia) and pragmatic factors. Receptive language is relatively better preserved than speech. The typically prominent smiling is a major signal in social interaction. Facial expression, bodily gestures, non-verbal vocalisation and touching are the preferred expressive modes, commonly within a limited repertoire. They serve mostly to indicate the patients' desires. Communication is mainly regulated by social partners rather than by the individual with Angelman syndrome. Communication ability may be enhanced through optimised interaction contexts with increased stability and visual interaction, facilitation of interaction with people and objects, facilitation of turn-taking and the use of consensual signals that may include spontaneous or learned gestures, pictograms or other communication devices.



# 10 MOVEMENT CONTROL

*Bernard Dan and Guy Cher*

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