Head or Hands - AAC for Girls with Rett Syndrome

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Rett syndrome is a severe, mutiply-disabling condition which has been identified only in females. It occurs in all races and ethnic groups throughout the world at a prevalence rate of approximately 1:10,000 live female births. The syndrome was first described by Dr. Andreas Rett, an Austrian, in 1965, but it wasn't until 1983 that articles on the syndrome were published in English-language medical journals.

Girls with Rett Syndrome appear normal at birth and for the first six months or so thereafter. Their hand use, social responses and initiation are normal, and they hit their first milestones on time. Then skill development stops, they appear to lose interest in playing, and their muscle tone often becomes very low. Repetitive hand movements, especially hand washing or wringing movements, take over from the wide range of hand movements of the typical infant. Skills already acquired, such as walking or talking, regress or are lost altogether.

Typically the girls are left without speech and often with little voluntary movement when the deterioration stops around puberty. The characteristic features of the condition include repetitive stereotyped hand movements, apnea and hyperventilation, seizures, scoliosis, ataxia and apraxia affecting mobility, speech and even eye gaze. (Hunter, 1998)

While girls with Rett Syndrome may be misdiagnosed as having non-specific mental retardation, autism or cerebral palsy, the announcement of the discovery of a gene for Rett Syndrome in September 1999 will presumably eventually lead to both more frequent and more accurate diagnosis of the condition.

Until recently all writing about Rett Syndrome described the girls as having severe mental retardation associated with severely impaired speech and comprehension. In our experience with some 20 girls and women with Rett Syndrome, however, their receptive problems have never been as severe as their extraordinarily severe expressive problems, problems which make accurate intellectual assessment virtually impossible. What we do know is that those girls and women for whom an effective means of expression is found demonstrate the capacity to learn. As David Koppenhaver pointed out in his closing address at Dublin in 1998, if they are given access to education they have the potential to acquire academic skills, including literacy. Unfortunately, to date only a small percentage of girls with Rett Syndrome have been given even a simple means of communication. As Hunter (1998) says, "Too many people read the outdated literature and base their goals on Rett's 1983 paper which is titled "A progressive syndrome of autism, dementia, ataxia and loss of purposeful hand use in girls".

The main factors which have hindered the provision of AAC for girls with Rett Syndrome are:

1. The initial assumption that the degenerative process ended in early death. It is now recognised that the deterioration ceases about the start of puberty, and an average life expectancty of sixty is suggested.
2. Research into Rett Syndrome has focussed on finding the cause, with a view to cure or pre-

vention, rather than on helping the girls and women who are alive now.
3. The presumption of severe mental retardation.
4. The severe and unique neuro-motor problems of these girls.

"We now know that the enormity of their physical impairments is their greatest handicap, often overshadowing their ability to prove their knowledge and understanding." (Hunter, 1998)

Kim Johnson, one of the authors of this paper, has Rett Syndrome. Kim's story illustrates both some of the difficulties faced by girls and women with Rett Syndrome, and the possibility of academic achievement given a means of communication and a supportive family and educational environment.

Kim was brought to DEAL by her mother in 1987 when she was 23 years old. At that time she had been diagnosed as having severe intellectual disability, autism, cerebral palsy and epilepsy. She could walk unsteadily with assistance, she could pick up finger foods but not use a spoon, and she had no speech. Her face often lacked expression, giving an impression of incomprehension.

Initiating purposeful movement was difficult for Kim. Even when she was eating a meal she sometimes needed a physical prompt to reach for the next mouthful, though she enjoyed her food. Feeding herself aside, virtually the only spontaneous hand movement Kim had was a continuous washing motion in her lap, rubbing hand against hand. This hand wringing was a very dominant pattern. Kim's mother described how Kim would sometimes stop to wring her hands while feeding herself, even though she was clearly hungry and had her eyes fixed on the piece of food she'd been reaching for when the wringing intervened. On the rare occasions when she did attempt other hand movements she had a significant intention tremor.

Because of Kim's difficulty in using her hands the DEAL therapists gave her a headpointer, a long stick attached to a headband. With it she was able to select letters on a small keyboard and show, for the first time ever, that she had learnt something during her schooling. It was clear that Kim had severe initiation problems. When questioned about her lack of movement later, she spelt MOTIVATION IS PRESENT, BUT I CAN'T MOVE. Once Kim demonstrated her awareness the fact that she was not otherwise interacting with her world was more noticeable.

If you can move your eyes reliably you can select items on eye-pointing boards, but Kim seemed to have apraxia of gaze and her eyes would get stuck in one position. If you can work one switch reliably you can work a computer, but Kim could not work a switch reliably - she could not get her muscles started to perform the necessary movement in the time allowed. We experimented with different arrangements of switches, headpointers, and hand use, but at the end of the day we were unable to improve on the headpointer.

In 1989, when she was twenty-five, Kim was diagnosed as having Rett Syndome. In Kim's case the diagnosis could be made with certainty because Kim's parents were keen amateur film-makers. There was a full and detailed visual record of Kim's life, starting even before she had left the maternity hospital. When all the pieces of film showing Kim growing up were edited together the changes were clearly visible. The beautiful baby, the infant playing and babbling, the sociable one-year old watching other children playing (although with the knowledge of hindsight one could note the restricted hand movements, the start of hand wringing), the passive but still smiling two year old, the generally unsmiling older child whose hands never seem to be apart.

Towards the end of 1991 Kim's parents arranged for a psychologist to assess her. She used her headpointer to respond to questions from the Weschler Adult Intelligence Scale. Testing was extraordinarily slow, and in three sessions of two hours each she only completed three subtests. However, her responses were good and the psychologist inferred that her intellectual capacity was at least of average level. Kim's parents did everything in their power to enrich her life with music,

talking books, and excursions. In 1994 she started attending Adult Education courses at a college and completed her Victorian Certificate of Education in 1997. She is believed to be the first woman with Rett Syndrome ever to attain a formal educational qualification. (Crossley, 1997)

It was perhaps fortunate that Kim had not been diagnosed with Rett Syndrome before her first attendance at DEAL, or the negative implications of the diagnosis may have impacted on the interventions she was offered. As it was, our experiences with Kim have had a positive impact on our expectations of other clients with Rett Syndrome, and have encouraged our efforts to find communication strategies suited to each individual. We have discovered that what works for one girl or woman does not necessarily work for the others. Currently we have clients with Rett Syndrome successfully using headpointers, eye pointing, auditory scanning and independent and facilitated hand pointing to access communication aids of varying complexity, ranging from picture boards to Lightwriters.

Our experiences with the older girls and women have lead us to initiate a research project with younger girls aimed at preserving or regaining hand function. Use of tight fitting Lycra garments as dynamic splints, enhancing proprioception and proximal stability, is central to the project. Similar garments have produced significant functional gains in children with cerebral palsy. (Blair et al, 1995) This presentation will report on this research, as well as detailing the outcomes of all DEAL's clients with Rett Syndrome. Videotape of Kim and of girls wearing Lycra garments will be shown.

REFERENCES

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