Case Study: Deterioration, Autism, and Recovery in Two Siblings

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ABSTRACT
Two siblings whose functioning deteriorated in the second year of life met criteria for autism. They recovered after a form of behavior modification that was successful in a previous study. Follow-up of that study and of the siblings demonstrated that recovery was enduring. It is hypothesized that such therapy succeeds by modifying a still-plastic neural circuitry. J. Am. Acad. Child Adolesc. Psychiatry, 1995, 34, 2:232–237. Key Words: deterioration, autism, therapy, recovery.

This report is of two siblings whose functioning deteriorated in the second year of life, until they clearly met DSM-III-R (1987) criteria for autistic disorder and then, after intensive behavioral and language therapy, made dramatic recoveries and now are functioning normally. Both children were not functioning normally before their deterioration. The case is unique in that it is the first case since 1985 (see Kurita, 1988) to describe two siblings with autism characterized by a period of deterioration, and it is the first reported case in which two siblings recovered from autism.

There are currently two major subgroups of autistic children described in the literature, whose clinical course includes a distinct period of deterioration. One is a large subgroup characterized by a loss of speech, usually around 18 months of age (Kurita, 1985). The other subgroup was first described by Heller (1930). It is called “disintegrative disorder” in DSM-IV (1994).

Our siblings do not meet the criterion of an “apparently normal” development for at least the first 2 years of life. In our opinion, however, the literature on the fewer than 80 cases of disintegrative disorder (Volkmar, 1992) does not document with confidence a normal 2 or 3 years after birth. Therefore, we will discuss our two siblings in the general context of autistic disorder that includes a period of deterioration (AD-D).

CASE STUDIES
Anne-Marie and Michel have an older brother, Daniel, who has developed normally and is 14 months older than Anne-Marie, who is 21 months older than Michel. Before Daniel’s birth, Mrs. Maurice suffered a stillbirth and three early first trimester miscarriages attributed to T strain mycoplasma infection. There is no family history of language or cognitive problems.

Anne-Marie
Anne-Marie was born by cesarean section because of a breech presentation after an unremarkable pregnancy. Birth weight was 6 pounds, 7 ounces; her Apgar scores were 9 at 1 and 5 minutes. Development was generally normal for 15 months. There were no delays in motor milestones, social smile, or eye contact. Anne-Marie said “mama,” “dada,” and “bye” at 12 months and had a vocabulary of 10 words at 15 months. She often played reciprocally with family members and as a toddler excitedly greeted her father with, “Hi, Daddy,” and sought her mother out for attention and hugs.

There were, however, some concerns as between 6 and 10 months, Anne-Marie would sometimes isolate...
TABLE 1
Scores on the Vineland Adaptive Behavior Scales

<table>
<thead>
<tr>
<th>Anne-Marie: Chronological Age (mo)</th>
<th>22</th>
<th>30</th>
<th>39</th>
<th>68</th>
</tr>
</thead>
<tbody>
<tr>
<td>Communication</td>
<td>10</td>
<td>27</td>
<td>47</td>
<td>71</td>
</tr>
<tr>
<td>Daily Living Skills</td>
<td>16</td>
<td>29</td>
<td>47</td>
<td>82</td>
</tr>
<tr>
<td>Socialization</td>
<td>12</td>
<td>27</td>
<td>48</td>
<td>67</td>
</tr>
<tr>
<td>Motor Skills</td>
<td>17</td>
<td>33</td>
<td>43</td>
<td>65</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Michel: Chronological Age (mo)</th>
<th>18</th>
<th>25</th>
<th>33</th>
<th>39</th>
<th>47</th>
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<tbody>
<tr>
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<td>12</td>
<td>10</td>
<td>28</td>
<td>38</td>
<td>51</td>
</tr>
<tr>
<td>Daily Living Skills</td>
<td>17</td>
<td>18</td>
<td>23</td>
<td>33</td>
<td>45</td>
</tr>
<tr>
<td>Socialization</td>
<td>13</td>
<td>9</td>
<td>21</td>
<td>27</td>
<td>42</td>
</tr>
<tr>
<td>Motor Skills</td>
<td>19</td>
<td>20</td>
<td>26</td>
<td>44</td>
<td>56</td>
</tr>
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Note: Scores reflect level of functioning in months.

herself and have to be drawn into play. She often looked serious; her parents wished for her to become more outgoing. After the deterioration and diagnosis, her parents looked back and some other behaviors then took on added importance. Between 10 and 15 months, Anne-Marie would sometimes stare at toys while turning them, push food around her plate with the side of her pinky finger and touch the beak of her "Big Bird" doll to the animal pictures that bordered the back cover of her Golden Classic Books.

The deterioration began to be evident around 15 months, with irritability and crying. Then, when Mrs. Maurice took a 4-day trip, Anne-Marie clung to the babysitter and insisted on sitting in the same position. In the following months, there was increased crying and a decline into extreme social detachment. At 18 months of age, Anne-Marie became attached to a red shovel and had tantrums when it was removed. She stopped greeting her father and stopped speaking. Her time was increasingly spent in meaningless perseveration, staring into space or at her fingers for up to 7 minutes. Diagnosis was made sequentially by the three authors between 21 and 23 months. She met DSM-III-R criteria A1, 3, 5; B1, 2, and C1, 2, 3, and 5 for autistic disorder. The first of four administrations of the Vineland Adaptive Behavior Scales (Sparrow et al., 1984) by Dr. Cohen was done at 22 months (Table 1). Therapy began at 23 months and is described under "Therapy" and in the Appendix.

After this initiation of therapy, there was rapid improvement commingled with the appearance of new symptoms for a period of approximately 16 months. Words were quickly reacquired but there was extreme echoing. There was a brief period of self-injurious behavior and refusal to walk on a tile floor. Echoing decreased as Anne-Marie approached 30 months of age, but there was pronominal reversal. Relatedness gradually improved. She began to attend a normal nursery school at 30 months (Table 1: second Vineland) but required special help. At 39 and 40 months, she no longer met criteria for autistic disorder (during reevaluations with R.P. and I.C.) but there was slight echoing, a tendency to withdraw, and a sing-song quality to her voice (Table 1: third Vineland). Thereafter, she attended a normal day camp at the age of 4 years, 4 months, and needed some prompting in the first weeks to participate. She then completed normal kindergarten (Table 1: fourth Vineland) and first grade. School reports document advanced academic skills and good socialization. She has progressed well in second grade.

Michel

Pregnancy was remarkable for bleeding during the seventh month due to a marginal placenta previa. Michel was born vaginally weighing 8 pounds, 4 ounces. Early development was unremarkable. Michel smiled at 2 months, walked at 9 months, and said "mama" and "dada" at 1 year. He added approximately six to eight words within the next 6 months but only responded to several phrases, such as "come here," "sit down," and "bath time." Moreover, toe walking was noted and he showed diminished interest in people other than family. This behavior made his parents uneasy, given their experience with Anne-Marie. Therefore, they asked Dr. Cohen to examine Michel at 18 months, when they brought Anne-Marie for a follow-up visit. Dr. Cohen did a short evaluation and administered the Vineland (Table 1). He did not think that Michel was autistic but was concerned about delays in communication and socialization. Mr. and Mrs. Maurice were alarmed, and they asked their daughter's behavior and language therapists for their opinions. Both had limited contact with Michel but believed that he was not autistic.

Other signs soon appeared. Michel stopped learning words, became irritable, and started having tantrums. One day he was observed to run up and down the hall while visually tracking the top border of the walls. Then, at 24 months, while in a restaurant,
he suddenly became frantic and was inconsolable for approximately 30 minutes. After this episode, he stopped speaking and eye contact diminished. He avoided people, often had tantrums when approached, and twirled spontaneously. At 24 months, he met DSM-III-R criteria A1, 2, 3, 4, 5, B1, 2, 3 and C1 and 2 for autistic disorder, and within weeks began therapy (Table 1: second Vineland). At 26 months, he began hand flapping, clung to pieces of fabric, and peered at objects out of the corners of his eyes. Withdrawal generally increased but in therapy he responded to a simple request and approximated a new word, thereby ending his mutism. In the next few months, he added words and made some word combinations. Then, at the beginning of a car trip, when he was 30 months of age, he spontaneously called out his brother's name, fearing that Daniel was being left behind.

Michel began nursery school at 33 months of age (Table 1: third Vineland). Much of his speech was now echolalic. There was a mix of normal and handicapped children in his class. His mother's presence was required at school because of his tantrums, but when the tantrums subsided after several months he was able to attend school alone. The echoing diminished but there was pronoun reversal, poor articulation, and a monotone vocal quality. Relatedness improved. A Vineland was done at 39 months (Table 1). Michel began to attend a mainstream nursery school at 47 months of age. His teachers, unaware of his history, reported no problems. His speech improved but at 47 months there were still mild problems with articulation and intonation as well as brief hand flapping. Dr. Cohen's impression was that Michel no longer met the criteria for autistic disorder (Table 1: fifth Vineland). At 53 months, the nursery staff reported that he was sociable and progressing well academically. He would occasionally strike out at home and at school when frustrated. A speech/language evaluation at 62 months documented no echoing and a spontaneous, flexible, appropriate, and precocious use of language. Then, in a mainstream prekindergarten, he was 2 years above grade level in reading. There was some (but diminishing) difficulty in sharing. Kindergarten has proceeded well. Hand flapping has stopped. Michel's teacher reports precocious academic skills and friendly social behavior. Some affective volatility remains.

Medical Workups

Both children were neurologically intact. Routine blood tests, amino acid screening, and fragile X testing were recommended. (These tests were overlooked during the stressful time of Anne-Marie's diagnosis.) Michel was tested and all the results were negative.

Therapy

Background. Therapy for both children was based on the work of Lovaas (1981). In his study (1987), an experimental group of 19 autistic children, averaging 34.6 months of age, received 40 hours of individual behavior therapy per week for a period of 2 or more years. Their parents were also trained in therapy and supplemented treatment. Two control groups received 10 hours of individual behavior therapy a week. The prorated mental ages of the experimental and two control groups were 18.8, 17.1, and 17.6 months, respectively. There were some methodological problems. Assignments to the groups could not be randomized and the ages of the children in the groups at the onset of treatment and at follow-up were somewhat different. These issues were addressed in the literature (Baer, 1993; Lovaas et al., 1989; Schopler et al., 1989) and do not detract, in our opinion, significantly from the results.

The subjects were compared, at the ages of 6 and 7 years, on intellectual functioning and school placement. The mean IQs of the experimental and two control groups were 83.3, 52.2, and 57.5, respectively. Nine of the experimental group completed normal first-grade classes and were promoted into a normal second grade. Their mean IQ was 107, with a range of 94 to 120. Of the control groups, only one child completed a normal first grade and was promoted into a normal second grade.

A follow-up was done in 1984–1985 (McEachin et al., 1993), which compared the experimental group (mean age: 13 years) to the first control group (mean age: 10 years). The mean IQs of the experimental and control groups were 84.5 and 54.9, respectively. Eight of the nine children who were in normal class placement at age 7 remained there, with the other child requiring special education. Of the other 10 in the experimental group, 1 had attained regular class placement. The children also were rated by using several instruments for adaptive and social functioning. The experimental
group was clearly superior to the control group, and the 8 children who remained in regular class appeared to be functioning normally.

The Children's Therapy. Therapy for Anne-Marie began at 23 months of age and for Michel at 25 months of age. Socialization, verbal and nonverbal communication, play, and cognition were addressed in individual, highly structured behavior therapy. Each area was broken down into a multitude of discrete goals, the accomplishment of which would lead to more complex goals. Reinforcements were used. (See Maurice, 1993, pp. 331–359 for details of the children's therapy.) Trained behavioral therapists provided five 2-hour sessions weekly for Anne-Marie for 23 months. Because Michel's progress in therapy was slower, he received an average of approximately 25 hours weekly until half-day nursery school began at 33 months of age. Then, for 9 months, he had 10 to 20 hours of therapy weekly, and 6 to 10 hours weekly over the next year until therapy was discontinued at 53 months of age. Mrs. Maurice was trained as a therapist, so therapy was continuous. Both children had thrice-weekly speech/language therapy.

DISCUSSION

It is surprising that more cases of AD-D in siblings have not been reported, given that AD-D comprises a large subgroup and the incidence of autism in siblings is 2% to 3% (Young et al., 1989). One could consider in our siblings some genetic component given the similarities in the progression of deterioration and recovery. However, there were also dissimilarities.

Our siblings join the nine patients of Lovaas who recovered with intensive behavioral therapy. Lovaas (1987) defined recovery as attaining a normal IQ and regular class placement. He recognized that, recovery thus defined, residual symptoms could be missed. Therefore, an expanded evaluation was done in the follow-up study. We suggest that recovery refer to individuals who once clearly met the criteria for autistic disorder and no longer meet criteria for any pervasive developmental disorder, nor have any eccentricities of behavior or language that clearly separate them from their peers. It seems that the symptoms of autism can leave their mark on the personalities of children recovered from autism in the form of personality traits, tendencies, or vulnerabilities. Anne-Marie's occasional quiet and reserve, and Michel's tendency to overreact when frustrated, are not any more pronounced than similar traits occurring in children who were never autistic.

The most recent school and parental reports continue to support our impression that the children have recovered. Moreover, there have been two recent professional observations of them. The primary behavioral therapist made a social visit to the family when the children were, respectively, 7 years, 9 months old, and 6 years old. Informal observation of the children lasted 2 hours and no residua of autism were noted (B. Taylor, personal communication). Four months after this visit, the children's parents provided to the authors a recently recorded 24-minute videotape of their three children during play. The children had previously constructed a cardboard spaceship and were taped as they played the crew on a mission. There was much activity, dialogue, and interaction during the play sequence. No social, communication, or behavioral residua of autism were noted. These observations could be faulted as being unstructured but have the advantage of being naturalistic, capturing the children when they were relaxed and spontaneous.

One could question whether the children's outcome could be attributed to anything but their treatment. Could they have recovered spontaneously? This is highly unlikely. They had deteriorated for 7 to 8 months without improvement before therapeutic intervention. Moreover, a literature search of the past decade revealed no papers on spontaneous recovery in autism. One could also question whether the children might have recovered if treated otherwise. This is unknowable but also unlikely. There are few prospective follow-up studies of preschool autistic children. Lord and Schopler (1989) and Freeman et al. (1991) followed young autistic children undergoing unspecified treatment for periods averaging 4½ and 12 years, respectively. Despite some improvement in cognitive functioning, there remained much impairment. There are reports of success from other early-intervention programs for autistic children, but their success is much more modest than that of Lovaas. These programs are similar to that of Lovaas in that they begin early, are structured and behavioral, and closely engage parents. Lovaas' program appears to be more intensive (Olley et al., 1993).

How it happens that some autistic children can, with therapy, recover from autism is as puzzling as
why those with AD-D deteriorate in the first place. The reviews of children with AD-D (Kurita, 1985, 1988; Volkmar, 1992) note variable medical workups and most often nonconclusive findings to explain the deteriorations. Future technology may clarify underlying changes in the neural substrates of children with AD-D.

The brain is in a state of maturational flux before and after birth. Prenatal neurogenesis and neuronal migration and postnatal elimination of excess neurons, axons, and synapses can be influenced by genetic or extrinsic factors (Rakic, 1991a) which could result in autistic deterioration.

These same maturational changes may also underlie brain plasticity and recovery from autism. Rakic (1991b) posits that “cell death and axon degeneration provides a basis for cortical developmental plasticity during at least the early portion of infancy” (p. 156). Kandel and Jessell (1991) review work that demonstrates how experience shapes the brain’s neural circuitry at critical stages of development. Kandel (1991) then provides a cellular model showing how experience fine tunes synapses during critical periods of development. After the critical periods, which are of as yet unknown duration, “existing connections become stable and much less susceptible to modification” (Kandel and Jessell, 1991, p. 957). One can hypothesize that in those autistic children whose condition is as yet modifiable, rigorous behavioral therapy modifies the neural circuitry before the condition becomes permanent (Cothen, 1994). The number of programs providing intensive behavior modification is increasing, and one of the authors (I.C.) knows of several other recoveries. These programs should generate more data about their efficacy and any predictors of success or failure. That our siblings began treatment approximately 1 year earlier than those in Lovass’ group and after periods of deterioration, raises questions about the optimal period in which to commence therapy and whether a period of deterioration is a predictor of success.

**APPENDIX**

The therapy provided to the children is called "applied behavioral analysis" and it essentially an intensive, highly structured form of behavior modification. It is done one to one; uses reinforcers such as food, praise, smiles, or tokens; and requires extensive data collection to monitor progress. It is done by trained therapists/teachers and requires that the child’s parents be trained so that therapy is continuous.

The first step in setting up a therapy program is careful observation of the child and delineation of his or her areas of developmental delay and/or deviancy. Relatedness, language, play, emotional response, self-help skills, and cognition may all require intervention. In each area, a hierarchy of objectives is established. Each objective is addressed in repeated trials until mastered, at which point a new objective is presented. The current objectives in each area of development are worked on throughout the day.

Autistic children often have difficulty attending to tasks. Before learning can occur, therapists and parents need the child’s attention. Looking at the therapist and being quiet are reinforced. Inattentiveness is consistently discouraged.

The behavioral techniques used include prompting, modeling, and generalization. For example, the therapist might prompt the child to label a chair by pointing to the chair while saying “ch.” After the child learns to label the chair with the prompt, the therapist tries to fade the prompt so that the child labels the chair without such help. In modeling, the therapist performs an action that the child is verbally directed to perform. For example, the therapist might model sitting down as an aid to teaching the child to respond to the request to sit down. After the child learns to respond to the request with the modeling, the modeling will be faded. Teaching the child to then sit in different chairs in different settings is generalization.

For more detail, the reader is referred to The Me Book by Lovass (1981) and Appendix 2 of Mrs. Maurice’s book (1993) in which she provides an overview of Michel’s therapy.

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DETERIORATION, AUTISM, AND RECOVERY IN SIBLINGS


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