Effects of Environmental Events on Smiling and Laughing Behavior in Angelman Syndrome

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Abstract

Angelman syndrome is a neurogenetic disorder associated with unique behaviors and characteristics, including an unusually happy expression, inability to speak, ataxia, mental retardation, and abnormal EEG. Previous research has suggested that smiling and laughing behaviors in Angelman syndrome are inappropriate, excessive, and dissociated from contextual events. In the present study, the variability of smiling and laughing behaviors of 3 individuals with Angelman syndrome was examined across typical social contexts. Results indicate that laughing and smiling increased during social situations and occurred at low levels during non-social situations. The behaviors, therefore, did not occur totally inappropriately, as has been suggested. The findings illustrate the need to divert attention to the examination of environmental influences on purported phenotypic behavior in genetic syndromes.

Angelman syndrome is a genetic disorder associated with a de novo deletion of chromosome 15q11–13 of maternal origin or with paternal uniparental disomy (Clayton-Smith, 1992; Magenis, Brown, Lacy, Budden, & LaFranchi, 1987). The syndrome is characterized by abnormalities or impairments in neurological, motor, and intellectual functioning. Clinical features include developmental delay, stiff or tremulous motor movements, gait ataxia, balance disorder, uplifted arms, epilepsy, and hypopigmented skin (Angelman, 1965). Individuals with Angelman syndrome have a reasonably typical orofacial structure, including microbrachycephaly, a wide mouth, tongue protrusion, widely or irregularly spaced teeth and a pointed chin (Williams, Gray, Hendrickson, Stone, & Cantu, 1989). electroencephalogram abnormalities have been reported in up to 80% to 90% of individuals with the syndrome (Zori et al., 1992).

The existing evidence from an amalgamation of case studies and reviews suggests that many individuals with Angelman syndrome have problems with eating and sleeping, are overactive and restless, have a short attention span, and engage in repetitive and stereotyped behaviors (Buntinx et al., 1995; Clarke & Marston, 2000). Excessive laughter, reported in over 80% of cases (Summers, Allison, Lynch, & Sandler, 1995) is perhaps the most salient and frequently cited behavioral characteristic of the syndrome. The existing literature proposes that the increased occurrence of laughing in Angelman syndrome may be linked to a more generalized social deficit characterizing the syndrome (Joleff & Ryan, 1993). Some investigators have suggested that laughter in Angelman syndrome has a low threshold for occurrence and that the outbursts tend to be relatively frequent and dissociated from contextual events (Bower & Jeavons, 1967; Élan, 1975). Other reports have described laughing in Angelman syndrome as "incessant," occurring without a stimulus, and with no response to environmental surroundings or emotional feelings (Nirenberg, 1991). Although there are conditions, such as gelastic epilepsy, which trigger laughing behavior as part of a convulsion, laughter does not appear to be related to seizure activity in Angelman syndrome (Clayton-Smith, 1992).

The suggestions made by researchers have not, as yet, led to an explanation as to why laughter is so frequent among individuals with Angelman syndrome. Even in persons without developmental disabilities, it is not fully understood as a psychosomatic behavior. Relatively little is known about emotion–behavior–cognition sequences, and the literature contains only a very partial account of the complex ways in which emotion is embedded within, influences, and is influenced by cognitions and behaviors (Johnson-Laird & Oatly, 2000). There is even some debate, for example, as to whether facial expressions (e.g., smiling and laughing) are, in fact, accurate indicators of emotion. Research by Ekman and his colleagues (e.g., Ekman, Friesen, & Ancoli, 1980) has shown that participants who watched film clips designed to evoke positive and negative emotions showed facial expressions that were congruent with the emotional content of the stimuli. However, when individuals were studied in more naturalistic "emotional" settings (Camras, Malatesta, & Izard, 1991; Fernandez-Dols & Ruiz-Belda, 1997), they tended not to smile when experiencing positive emotions but, rather, appeared to smile only when the situation involved a social interaction. This suggests that smiling may be determined more by context-specific social motives than by emotional experiences (Fridlund, 1994).

Despite the expressed interest in the characteristic "happy" disposition seen in individuals with Angelman syndrome, there have been no studies in which investigators have attempted to further specify or systematically examine the prevalent smiling and laughing behaviors. As is the case in most phenotypic work, attention has been primarily limited to identifying global behavioral patterns of the syndrome (Dykens & Rosner, 1999) and has been biased towards the underlying biological basis of the disorder, with little success in explaining the nature of the behaviors per se. The need to partially divert attention from purely genetically oriented research to the testing and documenting of environmental influences on prevalent behavioral manifestations in genetic syndromes has previously been highlighted by a number of studies focused on the social reinforcement of behaviors with a genetic or organic etiology. For example, Anderson, Dancis, and Alpert (1978) demonstrated that self-injurious behavior (SIB), shown by all individuals with Lesch-Nyhan syndrome, could be influenced by social–environmental factors in some cases. Similarly, Oliver, Murphy, and Crayton (1993) demonstrated that the stereotypic behaviors of a child with Rett syndrome could be influenced by different environmental contexts. Thus, although most researchers at present mainly consider genetic predisposition and neurochemical disturbance in genetic syndromes (Oliver et al., 1993), it is equally important to examine behavioral manifestations in relation to social and environmental influences.

In the current study our purpose was to examine environmental influences on one facet of the behavioral phenotype of Angelman syndrome by assessing the occurrence of smiling and laughing behavior in relation to social stimuli. In order to examine the interaction between environment and behavior, we exposed individuals participating in the study to several conditions in which social–environmental events were systematically manipulated. The rationale for employing such conditions was to examine the behavior of the children during typical social situations and, consequently, to determine whether it was affected by these conditions.

Method

Participants and Settings

Three children with Angelman syndrome were identified through medical contacts in England and Greece. Rebecca was a 7-year-old girl who had been diagnosed with Angelman syndrome and severe developmental disabilities. On the Vineland Adaptive Behavior Scale (Sparrow, Balla, & Cicchetti, 1984), she attained an age equivalent of 14 months. Rebecca attended a school for children with developmental disabilities in England, but the individual sessions for this study were conducted during the summer holiday period at her home in Greece. Rebecca did not speak but primarily used nonlinguistic means of expression, such as pointing, waving her hands, or stamping her feet. She was able to stand and walk unsupported, but her movements were stiff and jerky. Angelman syndrome was diagnosed when she was 6 years of age, following a display of the 15q deletion at molecular genetic analysis.

Maria, an 11-year-old girl diagnosed with Angelman syndrome and developmental disabilities, attained an age equivalent of 15 months on the Vineland Adaptive Behavior Scales. She attended a local school for children with developmental and behavioral disabilities in England. Maria had no speech but babbled frequently and made click-

ing, sucking, kissing, and gurgling noises. Like Rebecca, she used nonlinguistic means of expression, such as pointing or waving her hands, and was able to stand and walk unsupported. She was diagnosed as having Angelman syndrome at the age of 3.5 years, having displayed a deletion on chromosome 15q11–13. The experimental sessions for the current study were carried out at home in England.

Christopher, a 17-year-old male with Angelman syndrome, lived at home in Greece and attended a local school for children with special needs on a part-time basis. On the Vineland Adaptive Behavior Scales, he attained an age equivalent of 20 months. He had no speech, but communicated with his parents primarily through use of his personal sign language. Christopher could silently read 4- to 5-letter words and name pictures of animals. He could stand and walk with support; he had had two operations to his lower limbs due to walking difficulties. He was diagnosed with Angelman syndrome at the age of 15 years, following the display of the 15q 11–13 chromosomal deletion. All sessions were carried out at his home.

Conditions and Response Definitions

Each child was repeatedly exposed to three conditions. In the interaction condition, the experimenter (the second author) sat beside the child and provided continuous "enthusiastic" social interaction while playing with one of the child's favorite toys: plastic rings or toy animals, squeaky rubber toys, and the pieces of a puzzle illustrating an animal farmyard. In the proximity condition, the experimenter sat close to the child but did not initiate or engage in any sort of social interaction, even when attempts were made by the child to get the experimenter's attention. Toys provided were the same as just noted. In the alone condition (Rebecca and Maria only), the child was left alone, except for the presence of the camera operator and was allowed to engage in solitary activities, such as playing with a plastic toy, flicking through the pages of a magazine, or simply sitting in a chair. In the instructions condition (Christopher only), Christopher's mother engaged in table-top tasks with him; she used an instructive and authoritative tone of voice. The interaction revolved around activities that were often done at home, such as creating figures with play-dough or reading a picture book.

Blocks of three sessions were conducted daily, with eight blocks of sessions conducted for Rebecca and seven for Maria and Christopher. Within each block, the three conditions were presented in a random order, determined before the experiment began. Each condition lasted the duration of the time it took to get a full 5 minutes of unconcealed footage of the child's face and expressions (this usually involved 5 to 7 minutes of footage). Thus, if the child was observed to be mouthing an object in such a way that prevented observation of his or her face, these data were excluded from the analysis. A camera operator was in the room for all conditions but did not interact with the children. There was a break of approximately 5 to10 minutes between conditions.

Measurement and Interobserver Reliability

Two child behaviors were recorded. *Smiling* was defined as any horizontal stretching of the lips and an upturning of the corners of the mouth, frequently followed by the parting of the lips and viewing of the teeth. *Laughing* was defined as any short burst of inarticulate voiced noises, accompanied by an opening of the mouth, upturned corners of the lips, displayed teeth, half- or completely shut eyes, and raised cheeks. Observers collected data on response frequency and duration using an Olivetti Quaderno laptop PC. The software (Repp, Harman, Felce, Acker, & Karsh, 1989) allows up to 43 separate behaviors to be simultaneously recorded in continuous time and, thus, avoids any form of time-sampling.

Interobserver agreement was assessed by having a second observer simultaneously but independently collect data during 25% of sessions for Rebecca and 28.6% of sessions for Maria and Christopher. Kappa indices were calculated based on a 1-second interval-by-interval comparison of observer records. For smiling behavior, Kappa indices were .86, .70, and .80 for Rebecca, Maria, and Christopher, respectively. For laughing behavior, Kappa indices were .90, .60, and .93. All indices were equal to or greater than .6, suggesting that interobserver reliability was good.

Results

Figure 1 shows the percentage of time during which the children engaged in smiling and laughing in each session in each condition. Table 1 shows the corresponding means and standard deviations (*SD*s) for each child for each condition.

The figure shows that the percentage of time

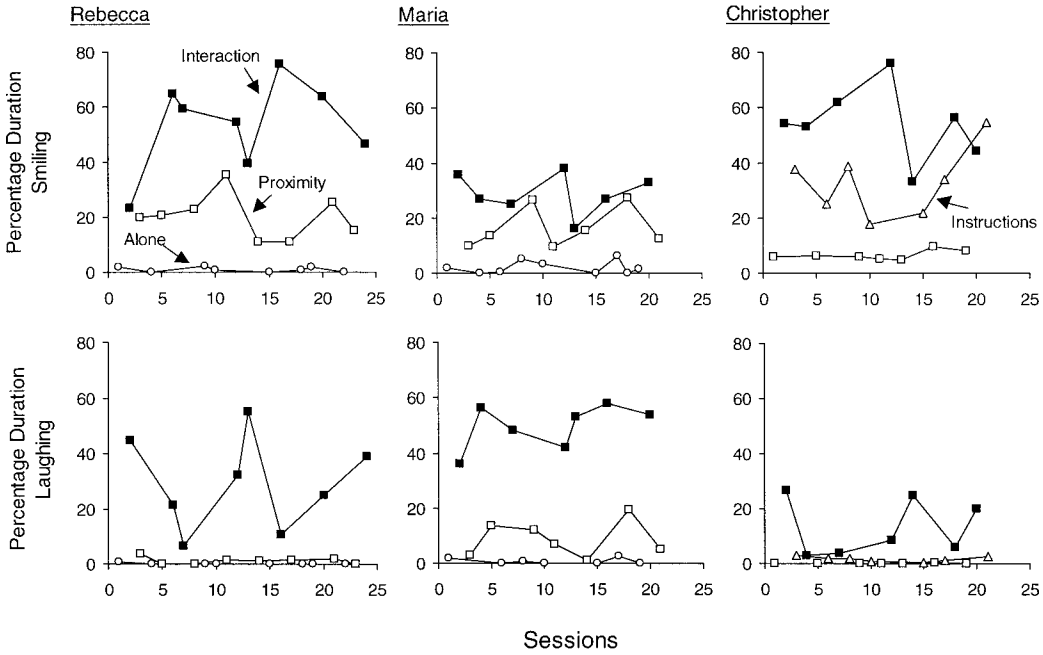

Figure 1. Percentage duration of laughing and smiling observed in each session in each condition for each child.

during which the children were engaged in smiling and laughing varied systematically across conditions. For Rebecca and Maria, high levels of smiling occurred in the interaction condition (53.39% and 28.81%, respectively), intermediate levels occurred in the proximity condition (20.20% and 16.39%), and low levels in the alone condition (.86% and 2.09%). For Christopher, high levels of smiling occurred in the interaction and instructions conditions (54.19% and 37.77% respectively), whereas low levels of smiling occurred in the proximity condition (6.50%).

Table 1. Mean Percentage Duration (and *SD*) of Smiling and Laughing Behavior Observed by Child and Condition

	Laughing		Smiling	
Child/Condition	Mean	*SD*	Mean	*SD*
Rebecca				
Alone	.08	.23	.86	.89
Interaction	29.27	16.63	53.39	16.52
Proximity	1.17	1.25	20.20	8.10
Maria				
Alone	.74	1.08	2.09	2.39
Interaction	49.63	8.02	28.81	7.40
Proximity	8.80	6.59	16.39	7.42
Christopher				
Instructions	1.63	1.00	32.77	12.51
Interaction	13.24	10.35	54.19	13.45
Proximity	.05	.12	6.50	1.76

As with smiling behavior, the degree to which the children engaged in laughing also varied across conditions. For Rebecca, high levels of laughing occurred in the interaction condition (29.27%), whereas low levels occurred in the proximity and alone conditions (1.17% and .08%, respectively). For Maria, high levels occurred in the interaction condition (49.63%), intermediate levels occurred in the proximity condition (8.80%), whereas low levels occurred in the alone condition (.74%). For Christopher, high levels of laughing were observed in the interaction condition (13.24%), and low levels were observed in the instructions and proximity conditions (1.63% and .05%, respectively).

For Rebecca and Maria, then, low levels of laughing and smiling occurred in the alone condition and intermediate levels, in the proximity condition. High levels of smiling and laughing occurred in the interaction condition. These data suggest that laughing and smiling occurred during contexts that were appropriate (i.e., those that involved social interaction). High levels of laughing and smiling did not occur in the alone and prox-

imity conditions. For Christopher, low levels of smiling and laughing behavior occurred in the proximity condition, intermediate levels in the instructions condition, and high levels of smiling and laughing behavior in the interaction condition. Again, these data suggest that laughing and smiling occurred in appropriate contexts.

Taken together, these data suggest that although the smiling and laughing behavior of these children was somewhat excessive (occurring for almost half of the time in the interaction condition for Maria, for example), the behaviors, nevertheless, occurred in a social and more appropriate context. Laughing and smiling was highest in the condition in which there was social interaction, as would be expected during this type of context, and the behaviors rarely occurred at times when the children were left alone.

Discussion

Previous research has repeatedly documented the increased incidence of smiling and laughing behavior in individuals with Angelman syndrome, with over 80% of this population (Summers et al., 1995) having been reported to exhibit "excessive" laughing behavior (Angelman, 1965; Buntinx et al., 1995). Individuals with Angelman syndrome are described as having a happy demeanor, being overexcitable, and exhibiting inappropriate bursts of laughter following minimal provocation. Interestingly, researchers' reports about the prevalence of such behavior do not appear to be substantially different from data obtained from parent-rating scores (Summers et al., 1995). This consistency indicates the salience and distinctiveness of such behavior and contributes to the delineation of the behavioral phenotype of this genetic syndrome. However, the frequency of the observation of smiling and laughing behavior has resulted in a somewhat stereotypic view of people with Angelman syndrome. Previous investigators have described laughter in Angelman syndrome as pathological (Bower & Jeavons, 1967; Robb, Pohl, Baraitser, Wilson, & Brett, 1989). These studies are useful, but they share one common limitation, namely, that the analysis reported is descriptive rather than experimental.

In the current study, the data show that smiling and laughing in children with Angelman syndrome appears to be influenced by social and environmental concomitants. Persistent close-to-zero occurrences of both behaviors in the alone condition contradict previous suggestions of the absence of any environmental antecedents for these behaviors (Elian, 1975). In the absence of any social interaction, smiling and laughing were manifested at a minimal level. In contrast, during the interaction condition, heightened levels of smiling and laughing behavior were recorded for all 3 children. These data implicate a conditional and predictable basis to smiling and laughing behavior in Angelman syndrome. The children's limited smiling and their lack of laughter during the proximity condition indicated that their behavior was affected by the presence of the experimenter, but less than in the interaction condition, where the manifestation of such behaviors was prevalent. We note that in the instructions condition, which was employed for Christopher only, it was not possible to determine whether the levels of smiling and laughing observed were affected by the individual with whom he was interacting (experimenter vs. mother) or type of social interaction (unconstrained vs. instructional). However, taken together, the differential data gathered from the children's responses to the conditions confirm that highly stereotypic behaviors associated with Angelman syndrome, namely, smiling and laughing, can be influenced by environmental and social factors.

The present study is, to our knowledge, the first experimental demonstration that laughing and smiling behavior in individuals with Angelman syndrome is dependent on contextual events. These data reconfirm that excess or problem behaviors can be affected by environmental situations, even when they may be biologically predetermined. Social and environmental factors have repeatedly been found to elicit and influence excess or problem behaviors that have organic or genetic antecedents. Carr and McDowell (1980) showed that the self-injurious scratching exhibited by a 10-year-old child, initially thought to be evoked by organic factors (contact dermatitis), subsequently began to be influenced by social reinforcement. The rate of scratching was positively correlated with the rate of social attention and could be systematically increased or decreased by the respective presentation or withdrawal of contingent social reinforcement. These findings illustrate the need to diverge from purely genetic research towards a systematic examination of the biological–environmental interaction.

The within-syndrome approach that we adopted in this study cannot be used to explain

whether the prevalence of the smiling and laughing behaviors among individuals with Angelman syndrome is specifically related to the syndrome or would be commonly reported among individuals of similar developmental levels but lacking the particular underlying genetic abnormality. Across-syndrome comparisons with a control group of individuals matched for age, gender, and severity of developmental disability may demonstrate quantitative and qualitative differences in seemingly shared behaviors (Dykens, 1995). Thus, syndromes that show partial specificity with Angelman syndrome may contribute to distinguishing between smiling and laughing behavior as an indicator of social interaction and such behavior as an unintentional and inappropriate consequence of genetic disorder. For example, individuals with Williams syndrome have been described as overactive, restless, stubborn, friendly, pleasant, affectionate, and very sociable (Dilts, Morris, & Leonard, 1990). An assessment of the behavior that these individuals exhibit across different social and environmental contexts may allow for a comparative measure of smiling and laughing behavior across syndromes. Behavioral specificity is very important in the search for gene–brain–behavior relationships, and discovering these correlations is fundamental in phenotypic research (Dykens & Rosner, 1999). Qualitative analyses of facial expressions should also be conducted for individuals with Angelman and other syndromes to establish the extent to which their expressions are appropriate or excessive within specific contexts.

Overall, the results of this study suggest that there is a need for further research in delineating the relationship between genetic predisposition and the behavioral phenotype of Angelman syndrome. The prevalent smiling and laughing behaviors associated with the syndrome have been found, on the basis of the data presented, to have social–environmental antecedents. Interactions between features of the syndrome and social and environmental factors challenge the notion of a unitary, genetically determined behavior. Future investigators need to deviate from global to more refined behavioral descriptions and examinations of the behavioral manifestations of Angelman syndrome and other genetic syndromes.

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