Published in final edited form as:

J Dev Phys Disabil. 2012 August ; 24(4): 327–335. doi:10.1007/s10882-012-9272-6.

Assessment and Treatment of Self-Injurious Behavior Associated with Donnai-Barrow Syndrome

Henry Roane,

Upstate Medical University, Syracuse, NY, USA

Kelly Bouxsein, and

University of Nebraska Medical Center's Munroe-Meyer Institute, Omaha, NE, USA

Caitlin Fulton

University of Nebraska Medical Center's Munroe-Meyer Institute, Omaha, NE, USA

Henry Roane: roaneh@upstate.edu

Abstract

Donnai and Barrow (*American Journal of Medical Genetics*, 68, 441–444, 1993) reported multiple participants who shared a variety of specific physical and neurological anomalies. Relatively few cases have been reported since then and few of those have progressed to an age such that the developmental progression of the disorder can be ascertained. We describe one participant with Donnai-Barrow syndrome who engaged in repetitive self-injurious behavior (SIB), which heretofore has not been described in this population. Direct observations of the participant's SIB were conducted across a range of contexts, and the behavior was successfully decreased using a combination of procedures. This case is discussed within the context of the behavioral phenotype of Donnai-Barrow syndrome.

Keywords

Donnai-Barrow syndrome; Self-injurious behavior; Behavioral intervention

Donnai and Barrow (1993) described a syndrome which included diaphragmatic hernia, exomphalos, hypertelorism, agenesis of the corpus callosum, sensorineural deafness, and myopia. Since then, Donnai-Barrow syndrome (DBS) has been identified in 27 cases, of which 10 were diagnosed following a terminated pregnancies or death secondary to other complications (e.g., congenital malformations; Pober et al. 2009). Significant developmental delays were noted in approximately 87% of the surviving sample reviewed by Pober et al.

Self-injurious behavior (SIB) involves deliberate engagement in responses that cause harm to one's self. Common examples of SIB include head banging, self biting, eye poking, pica, and self hitting. Such behavior is relatively common among individuals with developmental disabilities, and increases in likelihood as the level of developmental impairments worsens

(Hyman et al. 1990; Oliver et al. 1987). In addition, SIB is fairly common among individuals who have underlying genetic conditions. For example, the behavior has been reported among individuals with Smith-Magenis syndrome, Prader-Willi syndrome, and Cornelia DeLange syndrome (Arron et al. 2011). In these cases, however, the behavior is associated with these disorders likely as a function of the associated intellectual disabilities; it is not a hallmark feature of the disorders (Chassaing et al. 2003; Schroeder 1991). In contrast, there are some genetic conditions that are specifically associated with the occurrence of SIB. For example Lesch-Nyhan syndrome is associated with specific topographies of SIB including lip and finger biting (Nyhan 2005).

To date, the phenotype of DBS has been defined by a combination of physical malformations and developmental delays. However, there have been no reported cases describing the behavioral phenotype of a surviving participant with DBS. Thus, the current report concerns a case in which a child exhibited common features of DBS and additional SIB, which was successfully treated.

Method

Participant and Setting

The participant, who was 5 years old at the time of this intervention, was the result of a 36-week pregnancy born to healthy parents (mother aged 19 years and father aged 21 years). A prenatal ultrasound scan noted that presence of agenesis of the corpus callosum, which was confirmed within the month following his birth via MRI. At birth a metaphase FISH test returned no abnormal chromosomal results. The participant weighed 2507 g at birth, with a length of 49 cm, and head circumference of 36 cm. He showed diaphragmatic hernia via x-ray which was repaired surgically following birth, orbital hypertelorism, hearing loss, and other facial deformities. All features were indicative for categorization as DBS, which was designated by the attending geneticist. The participant had been previously diagnosed with profound mental retardation and had a history of tonic-clonic seizures.

The participant was referred for clinical treatment of SIB, which had resulted in a detached retina prior to his participation in this analysis. Previous attempts at behavioral management had been unsuccessful at reducing the occurrence of his SIB. Various medications (i.e., fluoxetine, risperidone, lorazepam, and clonidine), often prescribed to address similar challenging behavior, were also unsuccessful for decreasing the participant's SIB. As a result, the participant was referred to an outpatient behavioral treatment program that specialized in the assessment and treatment of severe SIB.

All sessions were conducted in a fully padded room (4 m by 4 m) equipped with a one-way observation window. The room consisted of a table, chairs, and various session-related materials (e.g., toys, instructional items). The participant received services 5 days per week, for 1.5 h per day. All functional analysis and treatment sessions were 10 min in duration and each session was separated by an approximate 2-min break. All preference assessment sessions were 3 min in duration, also separated by a brief break.

Response Measurement and Interobserver Agreement

SIB was defined as contact of the participant's closed fist against the side of his head or near his eyes. During those sessions of the treatment analysis that included a response blocking component, SIB was recorded if the participant's hand made contact with the therapists hand/arm while the therapist was implementing the blocking procedure (described below). Across all sessions, frequency data were collected on each occurrence of SIB. These data were converted to a response rate (responses per min; rpm) for the purpose of data analysis. During the preference assessment and treatment analysis, data were also collected on the duration of item interaction (defined as contact between one both hands and a target item or looking at the therapist). These data were analyzed via conversion to a percentage of session measure (i.e., the duration of item interaction in seconds divided by the duration of the observation, multiplied by 100).

Data were collected by individuals who had received prior training in direct observation. All data were collected on laptop computers equipped with a specialized data collection program. Observers were seated behind a one-way observation window for all sessions and were able to listen to any session-related activities through an intercom system. During 38.3% of all sessions, a second observer simultaneously collected data to assess interobserver agreement (IOA). For SIB, IOA was calculated by partitioning each 10-min session into a series of 10-s intervals and comparing intervals that contained observer agreements on the occurrence or non-occurrence of behavior (defined as both observers recording the same frequency of responses in a given 10-s interval) by the total number of intervals, multiplied by 100. For item interaction, IOA was calculated by dividing the smaller duration of observed item interaction by the larger duration of observed item interaction, multiplied by 100. The average IOA for SIB was 83.0% (functional analysis), 93.1% (preference assessment), 83.4% (response blocking treatment analysis), and 85.8% (response effort treatment analysis). For item interaction, IOA averaged 96.4% (preference assessment), 80.9% (response blocking treatment analysis), and 86.7% (response effort treatment analysis).

Procedure

Functional Analysis—Upon his admission to the outpatient program, the participant's SIB was observed in a variety of controlled contexts based on the procedures described by Iwata et al. (1994). Each observation lasted 10 min and the observations were conducted in a random order in accordance with a multielement single-case experimental design. In the demand condition, a therapist continuously presented academic-type instructions to the participant using successive verbal, modeled, and physical prompts. Contingent upon the occurrence of SIB, the participant was provided with a 20-s break from instructions (i.e., any academic materials were removed and the therapist turned away from the participant). In the attention condition, the participant was given access to a low preference toy with which to interact (identified via a pre-assessment preference assessment; Fisher et al. 1992) while the therapist was engaged in an attention-diverted activity (e.g., doing paperwork). Contingent on the occurrence of SIB, the therapist provided 20 s of attention in the form of statements of concern. The ignore condition consisted of the therapist and the participant being in the room together, but the therapist provided no programmed consequences following the

occurrence of SIB. In the tangible condition, the therapist provided pre-session access to and subsequently withheld the participant's preferred item (identified via the same preference assessment noted above) unless SIB occurred, upon which the participant was given 20-s access to the preferred item. Finally, a toy play condition was conducted as a control condition. In this condition, the participant received continuous attention from the therapist (e.g., playful comments) and continuous access to his preferred item, and no instructions were presented. There were no programmed contingencies for SIB in this condition.

Preference Assessment—Following the functional analysis, a competing items preference assessment (Piazza et al. 1996) was conducted to compare rates of SIB across different items and types of therapist attention. Each item or type of attention as well as a no-item/no-attention control was delivered for 3 min, and the participant was exposed to each condition three times. During each exposure, the participant received continuous access to the target stimulus (or nothing in the control condition) while observers recorded the concurrent occurrence of SIB and item interaction (when appropriate). For example, in the vibration condition the participant had access to a vibrating ball for the duration of the 3-min observation, whereas in the continuous-attention condition the therapist interacted with the participant continuously for 3 min. The no-item/no-attention control condition was also included to assess the occurrence of SIB in the absence of any alternative source of stimulation. The results of the preference assessment were summed across exposures to yield the mean occurrence of SIB and item interaction for each condition.

Response Blocking Treatment Analyses—Based on the results of the functional analysis and preference assessment, an intervention was initiated to evaluate the effectiveness of access to environmental stimulation on the occurrence of SIB. All treatment sessions were 10 min in duration and were conducted in the same physical arrangement as described above (i.e., a clinician in the padded room with the participant and observers seated behind an observation window). In the enriched environment (EE) condition, the participant had continuous access to the toy that was associated with lowest levels of SIB in the preference assessment and continuous adult attention. The EE plus response blocking (EE+RB) condition was conducted in an identical manner except that the therapist held his/her hand within 20 cm of either side of the participant's head such that the participant could no longer make contact between his hand and his head. The relative effects of the EE and the EE+RB conditions were compared in a reversal (ABAB) single-case experimental design.

Response Effort Treatment Analysis—Following the response blocking treatment evaluation, we conducted a second procedure to assess the effects of a response effort manipulation on the occurrence of SIB. Specifically, we evaluated the use of wrist weights in conjunction with EE (Hanley et al. 1998; Van Houten 1993). Following a baseline which was identical to the EE condition described previously, we manipulated the response effort associated with SIB by placing a 1.4 kg wrist weight on the participant's right wrist, which was the dominant hand the participant used when engaging in SIB. The application of the weight did not preclude the occurrence of SIB in that the participant could lift his right hand with the weight on his wrist and he could engage in SIB with his left hand (though this

rarely occurred). With the exception of the wrist weight, this condition was identical to the EE condition described above. The effects of EE and EE with the wrist weights (EE+WW) were evaluated in a reversal (ABAB) single-case experimental design.

Results

The results of the functional analysis (top panel of Fig. 1) were inconclusive as the participant exhibited SIB across all conditions. However, he was noted to engage in less SIB in the toy play condition in which preferred alternative stimuli (i.e., favorite toys) and continuous adult attention were present. These results suggest two outcomes. First, the participant's SIB was not clearly influenced by a specific environmental context; a finding that is in accordance with SIB associated with other genetic conditions (Mace and Mauk 1995). Second, the lower levels of SIB observed in the presence of alternative stimuli indicated that the provision of alternative sources of stimulation (i.e., access to toys, interaction with a clinician) would reduce the occurrence of SIB.

Based on the outcome of the functional analysis, we conducted a preference assessment to identify stimuli that would compete with the occurrence of SIB (i.e., produce relatively high levels of interaction and relatively low levels of SIB). As depicted in the bottom panel of Fig. 1, the vibrating ball and therapist attention were associated with the lowest levels of SIB. These results led to the development of the EE condition as a potential intervention.

As shown in the top panel of Fig. 2, relatively high levels of SIB occurred during the initial exposure to the EE condition ($M=7.0$ rpm). Thus, an additional component was added to the treatment in which the therapist blocked all contact between the participant's hand and his face and eyes (EE+RB). As shown in the second phase on the top panel of Fig. 2, the EE +RB intervention resulted in an immediate and sustained reduction in SIB ($M=0.2$ rpm). To ensure the intervention was responsible for the observed change in the participant's SIB, the treatment was briefly withdrawn and the EE condition was re-implemented. SIB increased to previous levels in the return to the EE condition ($M=13.1$ rpm). Subsequently, the EE+RB condition was implemented and SIB again decreased to near-zero levels ($M=0.2$ rpm).

Although the EE+RB procedure appeared effective in reducing the occurrence of SIB, this procedure involved near-continuous monitoring of the participant along with response blocking, which may be of limited practicality. Thus, the effects of wrist weights in conjunction with EE were evaluated. These results are depicted in the bottom panel of Fig. 2. Variable levels of SIB occurred in the initial EE phase ($M=4.2$ rpm). When the weight was applied to the participant's right wrist (EE+WW), SIB decreased ($M=1.0$ rpm). This general pattern was replicated during the return to the EE ($M=4.8$ rpm) and EE+WW ($M=0.8$ rpm) conditions. It should be noted that the final phase of the EE+WW condition ended prematurely due to a change in the participant's living situation. Nevertheless, the results of the response effort treatment analysis suggest that the use of wrist weights was a viable alternative to the response blocking procedure.

Discussion

Previous descriptions of DBS have revealed the following characterization: diaphragmatic hernia, exomphalos, hypertelorism, agenesis of the corpus callosum, sensorineural deafness, and myopia (Chassaing et al. 2003; Donnai and Barrow 1993; Gripp et al. 1997; Pober et al. 2009). Although a primary developmental characteristic of DBS includes developmental delay (Pober et al.); to date, there has been no documentation of DBS being associated with the occurrence of SIB, though this behavior is relatively common among individuals with other genetic conditions (Arron et al. 2011) and developmental delays (Schroeder 1991).

The current participant displayed clinically significant SIB that had resulted in previous medical complications (i.e., detached retina). This behavior had negatively impacted the participant's physical health as well as his social and educational development (i.e., persistent SIB interfered with others interacting with him). The participant was responsive to behavioral intervention. The intervention consisted of providing the participant with response independent access to preferred stimuli, adult interaction, response blocking, and a response effort manipulation. Moreover, the intervention was effective at maintaining low levels of SIB and the participant was able to be promoted beyond his educational placement at the conclusion of service provision.

The current case may produce consideration toward revising the phenotype of DBS, particularly with regard to the behavioral characterization of this disorder. Although SIB has been noted as a phenotypic expression of other disorders (e.g., Nyhan 2005), the occurrence of SIB has not been documented within DBS. This may be due, in part, to the relative lack of developmental information on DBS participants. Thus, it may be the case that the underlying DBS genotype influences the phenotypic expression of SIB in this population (which may be relatively unknown given the lack of long-term developmental data for this population). Alternatively, it could be the case that the developmental disabilities associated with DBS predispose certain participants with this disorder to develop various topographies of SIB. This course of behavioral development would be similar to the profile of the emergence of SIB among individuals with severe to profound mental retardation. Further work is needed to delineate the relation between DBS and the occurrence of SIB. In addition, future investigations might examine the extent to which SIB shows similar topographical and functional characteristics to those observed in the current case.

Mace and Mauk (1995) described a bio-behavioral model of SIB, the logic of which was applied to the interpretation of the current functional analysis results. Specifically, Mace and Mauk suggested that patterns of responding during functional analysis could be categorized along three dimensions: operant, biological, or mixed. An operant categorization was suggested for functional analyses that are characterized by low levels of SIB in the toy play (control) condition and differentially high rates of SIB in one or more of the test conditions. SIB was suggested to be categorized as biological when functional analysis results were characterized by high and undifferentiated levels of SIB across all conditions. Finally, a mixed categorization was suggested for situations in which the functional analysis results were characterized by generally high levels of SIB across all conditions of the functional analysis but one or more test conditions revealing differentially higher levels of SIB.

The results of the current participant's functional analysis suggested that his behavior could be characterized as operant using the Mace and Mauk (1995) model. That is, the participant's functional analysis was associated with generally undifferentiated levels of SIB across all test conditions but lower levels in the toy play (control) condition. However, a reinforcement-based treatment (EE) was not successfully in producing sustained reductions in SIB. It was not until response blocking and later, wrist weights were introduced that SIB decreased to low levels. The nature of the response blocking procedure prevented the participant from making contact between his hand and his face/eyes, whereas the use of wrist weights increased the presumed effort associated with engaging in SIB. Thus, the reduction in SIB associated with these procedures and the ineffectual use of reinforcement alone suggest that the participant's SIB was influenced by both operant and biological variables. This hypothesis is speculative, however, as the function of the response blocking component is unknown (Lerman and Iwata 1996).

In sum, the life expectancy of individuals with relatively rare genetic conditions is increasing (Katz 2009; Kirby et al. 2010). One consequence of this is that researchers and clinicians will be able to learn more about the characteristics of previously understudied populations. A related consequence is that many practitioners may find themselves confronted with participants who present with a rare chromosomal disorder and who require additional therapies. The current case, along with other reports (e.g., Sloneem et al. 2011; Swingle et al. 2006) suggest that behavioral assessment tools and behavioral interventions may be helpful in identifying variables that lead to the remediation of behaviors associated with underlying genetic conditions.

References

- Arron K, Oliver C, Moss J, Berg K, Burbidge C. The prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes. *Journal of Intellectual Disability Research*. 2011; 55:109–120. jir. [PubMed: 20977515]
- Chassaing N, Lacombe D, Carles D, Calvas P, Saura R, Bieth E. Donnai-Barrow syndrome: Four additional participants. *American Journal of Medical Genetics*. 2003; 121A:258–262. [PubMed: 12923867]
- Donnai D, Barrow M. Diaphragmatic hernia, exomphalos, absent corpus callosum, hypertelorism, myopia, and sensorineural deafness: A newly recognized autosomal recessive disorder? *American Journal of Medical Genetics*. 1993; 47:679–682. [PubMed: 8266995]
- Fisher W, Piazza CC, Bowman LG, Hagopian LP, Owens JC, Slevin I. A comparison of two approaches for identifying reinforcers for persons with severe and profound disabilities. *Journal of Applied Behavior Analysis*. 1992; 25:491–498. [PubMed: 1634435]
- Gripp KW, Donnai D, Clericuzio C, McDonald-McGinn DM, Guttenberg M, Zackai EH. Diaphragmatic hernia-exomphalos-hypertelorism syndrome: A new case and further evidence of autosomal recessive inheritance. *American Journal of Medical Genetics*. 1997; 68:441–444. [PubMed: 9021018]
- Hanley GP, Piazza CC, Keeney KM, Blakeley-Smith AB, Worsdell AS. Effects of wrist weights on self-injurious and adaptive behaviors. *Journal of Applied Behavior Analysis*. 1998; 31:307–310. [PubMed: 9652108]
- Hyman SL, Fisher W, Mercugliano M, Cataldo MF. Children with self-injurious behavior. *Pediatrics*. 1990; 85:437–441. [PubMed: 2304806]
- Iwata BA, Dorsey MF, Slifer KJ, Bauman KE, Richman GS. Toward a functional analysis of self-injury. *Journal of Applied Behavior Analysis*. 1994; 27:197–209. Reprinted from *Analysis and Intervention in Developmental Disabilities*, 2, 3–20, 1982. [PubMed: 8063622]

- Katz RT. Are children with Cerebral Palsy and developmental disability living longer? *Journal of Developmental and Physical Disabilities*. 2009; 21:409–424.
- Kirby RS, Lane JB, Childers J, Skinner SA, Annese F, Barrish JO, et al. Longevity in Rett syndrome: Analysis of the North American database. *The Journal of Pediatrics*. 2010; 156:135–138. [PubMed: 19772971]
- Lerman DC, Iwata BA. A methodology for distinguishing between extinction and punishment effects associated with response blocking. *Journal of Applied Behavior Analysis*. 1996; 29:231–234. [PubMed: 8682737]
- Mace FC, Mauk JE. Bio-behavioral diagnosis and treatment of self-injury. *Mental Retardation and Developmental Disabilities Research Reviews*. 1995; 1:104–110.
- Nyhan WL. Lesch-Nyhan disease. *Journal of the History of the Neurosciences*. 2005; 14:1–10. [PubMed: 15804753]
- Oliver C, Murphy GH, Corbett JA. Self-injurious behavior in people with mental handicap: A total population study. *Journal of Mental Deficiency Research*. 1987; 31:147–162. [PubMed: 3625761]
- Piazza CC, Fisher WW, Hanley GP, Hilker K, Derby KM. A preliminary procedure for predicting the positive and negative effects of reinforcement-based procedures. *Journal of Applied Behavior Analysis*. 1996; 29:137–152. [PubMed: 8682733]
- Pober BR, Longoni M, Noonan KM. A review of Donnai-Barrow and Facio-Oculo-Acousticorenal (DB/FOAR) syndrome: Clinical features and differential diagnosis. *Birth Defects Research*. 2009; 85:905–913. [PubMed: 19760683]
- Schroeder, SR. Self-injury and stereotypy. In: Matson, JL.; Mulick, JA., editors. *Handbook of Mental Retardation*. Elmsford: Pergamon; 1991. p. 382-396.
- Sloneem J, Oliver C, Udwin O, Woodcock KA. Prevalence, phenomenology, aetiology and predictors of challenging behaviour in Smith-Magenis syndrome. *Journal of Intellectual Disability Research*. 2011; 55:138–151. [PubMed: 21199049]
- Swingle HM, Ringdahl JE, Mraz R, Patil S, Keppler-Noreuil K. Behavioral management of a long-term survivor with tetrasomy 18p. *American Journal of Medical Genetics*. 2006; 140A:276–280. [PubMed: 16411237]
- Van Houten R. The use of wrist weights to reduce self-injury maintained by sensory reinforcement. *Journal of Applied Behavior Analysis*. 1993; 26:197–203. [PubMed: 8331016]

Fig. 1. Rates of SIB across conditions of the functional analysis (*top panel*). Mean percentage of session in which item interaction (grey bars) and SIB occurred during the competing items preference assessment (*bottom panel*)

Fig. 2. Rate of SIB during the Enriched Environment (EE) plus Response Blocking (RB) treatment analysis (*top panel*) and during the EE plus Wrist Weights (WW) treatment analysis (*bottom panel*)