Developmental Neurorehabilitation

ISSN: 1751-8423 (Print) 1751-8431 (Online) Journal homepage: https://www.tandfonline.com/loi/ipdr20

Toilet training in individuals with Angelman syndrome: A case series

Maartje Radstaake, Robert Didden, Nienke Peters-Scheffers, Dennis W. Moore, Angelika Anderson & Leopold M. G. Curfs

To cite this article: Maartje Radstaake, Robert Didden, Nienke Peters-Scheffers, Dennis W. Moore, Angelika Anderson & Leopold M. G. Curfs (2014) Toilet training in individuals with Angelman syndrome: A case series, Developmental Neurorehabilitation, 17:4, 243-250, DOI: 10.3109/17518423.2013.783140

To link to this article: https://doi.org/10.3109/17518423.2013.783140

Published online: 19 Aug 2013.

DEVELOPMENTAL NEUROREHABILITATION

http://informahealthcare.com/pdr
ISSN: 1751-8423 (print), 1751-8431 (electronic)

Dev Neurorehabil, 2014; 17(4): 243–250
 DOI: 10.3109/17518423.2013.783140

informa healthcare

ORIGINAL ARTICLE

Toilet training in individuals with Angelman syndrome: A case series

Maartje Radstaake[1,2], Robert Didden[1], Nienke Peters-Scheffers[1,3], Dennis W. Moore[4], Angelika Anderson[4], & Leopold M. G. Curfs[5]

[1]Department of Special Education, Radboud University Nijmegen, Nijmegen, The Netherlands, [2]Daelzicht, Heel, The Netherlands, [3]Driestroom, Nijmegen, The Netherlands, [4]Department of Education, Monash University, Melbourne, Australia, and [5]Department of Clinical Genetics, Maastricht University Medical Centre, Maastricht, The Netherlands

Abstract

Objective: To assess if adapted versions of the response restriction toilet training protocol, based on the behavioral phenotype of Angelman syndrome (AS), were successful in fostering urinary continence in seven individuals with AS.
Method: Data were collected in AB-designs during baseline, training, generalization and follow-up. The response restriction protocol was adapted: individuals were trained in their natural environment, were prompted to void and along with improving continence, the interval between voids was prolonged and time-on-toilet decreased.
Results: During generalization five individuals had less than two accidents and one to six correct voids per day; during baseline more accidents and/or less correct voids occurred. In two participants correct voids increased, but several accidents still occurred. Three participants maintained positive results after 3–18 months.
Conclusion: Despite their intellectual and behavioral challenges, urinary continence can be acquired in AS. Several indications of voiding dysfunctions were found; further research is indicated.

Keywords

Angelman syndrome, response restriction, toilet training

History

Received 4 March 2013
Accepted 5 March 2013
Published online 14 August 2013

Introduction

Angelman syndrome (AS) is a neurodevelopmental disorder caused by the absence of expression of maternally imprinted genes in the region at 15q11-13 [1]. Individuals with AS have severe intellectual disability (ID), motor and speech deficits, and epilepsy is often present [2]. In addition, they show jerky, tremulous movements and generally have an easily excited, happy demeanor. Incontinence is also common with individuals with AS, although prevalence rates differ across studies. For example, Buntinx et al. [3] found that 62.5% of a sample of children aged 2–16 years ($n = 23$) were incontinent for urine and feces during the day. Of individuals aged 16 and older ($n = 18$), 12.5% were incontinent. Another study found that, when sent to the toilet on a regular basis, 12 out of 28 adults (43%) with AS were incontinent for urine throughout the day [4].

Incontinence is associated with a range of adverse effects. Being incontinent may cause stigmatization, urinary tract infections, physical discomfort, and it may lead to dependency on caregivers and exclusion from certain activities or peer groups (see, e.g. [5, 6]). Further, it can place a burden on caregivers. Costs of diapers and medical treatments and wages for caretakers are also considerable [7].

Research on toilet training individuals with AS is needed to help address these issues. To our knowledge, the one published study addressing toilet training in AS used a modified Azrin–Foxx procedure to toilet train six individuals aged 6–19 years who were living in a residential facility [8]. The training mainly took place in the bathroom and included increased fluid intake, scheduled toileting, rewards for correct voiding, together with overcorrection and time out from reinforcement upon an incorrect void (i.e. accident). Positive practice was not used because of participants' motor limitations. Training resulted in increased correct voiding but, after training, accidents still occurred. Results were maintained over a period of two-and-a-half years for five out of six participants. The authors suggested that behavioral characteristic of individuals with AS (noncompliance, hyperactivity and bursts of laughter) may have impacted the efficacy of this intervention procedure, lengthening the training time required. This suggests that future toilet training procedures should be adapted to the behavioural characteristics of individuals with AS.

Duker, Averink and Melein [9] developed the response restriction (RR) method which differed from other procedures in that overcorrection was not used and bladder control was the aim of training, not self-initiated voiding. The RR procedure entails: (a) increased fluid intake, (b) a 10-min reinforcement interval in the bathroom with toys and praise upon correct voiding, (c) positive practice upon a urinary accident, (d) increasing the distance between participants

Correspondence: Maartje Radstaake, Department of Special Education, Radboud University Nijmegen, P.O. Box 9104, 6500 HE Nijmegen, The Netherlands. Tel: 0031-24-3612914. E-mail: m.radstaake@pwo.ru.nl

Dev Neurorehabil, 2014; 17(4): 243–250

and toilet seat following correct voids and (e) stepwise generalization to the living/daycare group once the training phase is completed. Training principally occurs in the bathroom where the participant has to stand near the toilet. Any behavior other than correct toileting (e.g. flushing the toilet, stereotypic behaviors, sitting on the ground) is prevented (i.e. restricted) by the trainer. Verbal interactions and eye contact between trainer and participant are avoided as much as possible and prompting is withheld.

The RR procedure has been shown to be effective in individuals with severe ID, but the effects of this protocol (with adaptations) with individuals with AS are unknown. In the present study, seven individuals with AS were toilet trained using a RR procedure which was modified to accommodate the aforementioned characteristics of individuals with AS. The aim of this study was to assess the effects of a modified RR procedure on urinary accidents and correct voids within a prompted toileting schedule.

Methods

Participants

Participants were recruited through the Dutch Angelman Foundation, an organization for parents who have a child with AS. Individuals were included if they were: (a) incontinent of urine throughout the day, (b) able to sit for 5 min, (c) showed regular voiding (wet diapers), (d) were able to walk independently, (e) followed simple instructions and (f) had no seizure activity.

Seven participants with a mean age of 14 years (range: 6–25) were included (see Table I). Five participants lived at home, while two lived in a residential facility. During daytime, all participants attended a daycare centre or school for individuals with ID. They were non-verbal and communicated through vocalizations and gestures. All parents gave their informed consent for their child's participation in this study.

Setting

Training was implemented in the setting where accidents most often occurred. Depending on the participant's voiding pattern, training took place in the bathroom of the participant's home or daycare center/school. Training occurred between 9 and 3 or 4 pm. The time the participant actually spent in the bathroom during the day, depended on the progress of the training (see Table II). When the child was not in the bathroom, s/he followed the normal curriculum in the class/daycare room or home and the attendance of the trainer in this period was slowly faded out.

Bathrooms included a toilet and were between 2 and 10 m^2 in size. Distracting objects (e.g. toilet brush, diapers) were removed and a table and one or two chairs were placed in the bathroom to seat the trainer(s). Peers were brought to another bathroom or toileting visits of the participants and peers were coordinated such that the toilet was available for training.

Table I. Participant characteristics.

	Sex (M/F)	Age (years)	Developmental age (years; months)	Subtype AS	Epilepsy
Mandy	F	6	1;6[b]	Imprinting error	No
Daniel	M	18	1;2–1;4[a]	Deletion	Yes
Ella	F	25	1;0–1;2[a]	Deletion	Yes
Nate	M	21	1;4[a]	Deletion	No
Alice	F	7	1;2–1;4[a]	Mutation	Yes
Jonah	M	8	1–1;2[a]	Deletion	No
Kevin	M	13	1;2[a]	Deletion	No

AS = Angelman syndrome.
[a]Score derived from the Vineland Adaptive Behavior Scales (VABS, [14]).
[b]Score derived from the Bayley Scale of Infant Development (BSID-II-NL, [15]).

Procedure

Each case study protocol in our training consisted of three consecutive phases: baseline, training and generalization. The baseline phase lasted four to five days, the training phase was completed when the participant had remained dry for 1.5 to 2 h during two to three consecutive reinforcement intervals following a correct void, after which the generalization phase was initiated, which lasted four to five days.

Prior to baseline, caretakers changed participants' diapers in the bathroom to familiarize them with the setting. During baseline diapers were removed and hourly toileting visits to the bathroom were scheduled, lasting for five minutes or until voiding occurred. No consequences were scheduled for correct and incorrect voids (see Table III). Also, parents and caretakers were asked which items their child or client preferred (following the preference assessment procedure by Sigafoos, Didden, & O'Reilly, [10]) and these items were placed on a small table in the bathroom, in sight of the participant, but out of reach.

The RR training protocol was adapted on several components (see Table IV), Additional and background information on the training components may be found in Duker et al. [9], Didden et al. [8] and Kroeger and Sorensen-Burnworth [6]. In Table II, the interventions per participant are outlined. Clarifications on the selection rationale for components per participant are available from the first author. Outlines for the baseline and generalization phase can be found in Table III.

This study focuses on urinary continence. When participants showed signs of defecating, they were brought to the toilet and verbally prompted to defecate in the toilet. When defecation occurred in the toilet, reinforcement was given similar to when a correct urinary void occurred. When they voided instead of defecating during this extra-training opportunity, they were reinforced according to the outlines in Tables II and IV. When this opportunity was less than 30 min prior to the following scheduled toileting time, 30 min were added to the following toileting visit.

Design

Data were collected in a case series design, wherein some components of the training differed per participant (see Table II). The number of correct voids and accidents during the baseline and generalization phases were individually assessed. During the training phase, all training components were implemented at once, while most training components were faded out in the generalization phase (see Table III). For Jonah, a second training was initiated after the first

Table II. Setting, training components, individual adaptations or additions and correct voids and accidents per participant.

Child	Setting	Used training components	Individual adaptations per training component or additions	Mean nr correct voids BS–GEN	Mean nr accidents BS–GEN
Mandy	School	Increased fluid intake	150 ml/30 min	0–2.3	0.5–0
		Response restriction			
		Toy play on toilet			
		Most to least prompting and praise			
		Increasing the reinforcement interval	30, 60, 75, 90 to 120 min after every second reinforcement interval without an accident.		
		Reinforcement interval in natural environment			
		Decrease in time-on-toilet			
		Remain seated on toilet following an accident			
Daniel	Living facility	Increased fluid intake	150 ml/30 min	1.7–3.8	1–0.5
		Response restriction			
		Most to least prompting and praise			
		Increasing the reinforcement interval	60, 75, 90 to 120 min after every second reinforcement interval without an accident.		
		Reinforcement interval in natural environment			
		Decrease in time-on-toilet			
		Remain seated on toilet following an accident			
Alice	Day care centre	Response restriction	Was seated for 5 min or until voiding occurred.	0–3.6	1.8–0.2
		Toy play on toilet			
		Most to least prompting and praise			
		Increasing the reinforcement interval	60, 75, 90 to 120 min after every second reinforcement interval without an accident.		
		Reinforcement interval in natural environment			
		Positive practice			
Jonah–1	Day care centre	Increased liquid intake	100 ml/hour	0.2–1.6	1.8–0.8
		Response restriction	Was seated for 10 min or until voiding occurred.		
		Toy play on toilet			
		Most to least prompting and praise			
		Increasing the reinforcement interval	60, 75, 90 to 120 min after every second reinforcement interval without an accident.		
		Reinforcement interval in natural environment			
		Positive practice			
Kevin	Day care centre	Response restriction	Kevin had to stand in front of the toilet, was only verbally prompted to urinate on the toilet.	0.2–2.6	1.6–0
		Increased fluid intake			
		Verbal prompting and praise			
		Reinforcement interval in natural environment	30, 60, 75, 90 to 120 min after every second reinforcement interval without an accident.		
		Increasing the reinforcement interval	Instead of response restriction, all his non-toileting behaviors were ignored.		
		Decrease in time-on-toilet			
		Positive practice			
Ella	Living facility	Increased fluid intake	200 ml/30 min, until she had drunk 2000 ml without urinating.	0–1.5	0–0.3
		Response restriction	Ella had to stand in front of the toilet seat, was only verbally prompted to urinate on the toilet.		
		Verbal prompting and praise	Distance to toilet seat was increased from 30 cm, 50 cm, 75 cm, 100 cm, 150 cm, 200 cm up to entire bathroom (8 m^2), after every consecutive correct void.		
		Reinforcement interval in natural environment	Reinforcement intervals were increased from; 10, 15, 30, 45, 60, 90 up to 120 min, when Ella had stayed dry between two voids.		
		Increasing the reinforcement interval	After an accident, Ella received a verbal reprimand and was taken to the toilet to see if she continued voiding.		

(*continued*)

Dev Neurorehabil, 2014; 17(4): 243–250

Table II. Continued

Child	Setting	Used training components	Individual adaptations per training component or additions	Mean nr correct voids BS–GEN	Mean nr accidents BS–GEN
Nate	Home	Increased fluid intake Response restriction Most to least prompting and praise Reinforcement interval in natural environment Increasing the reinforcement interval Decrease in time-on-toilet	400 ml/hour, until he had drunk 3000 ml. His mother, accompanied by a trainer, sat him down on the toilet seat for a maximum of 30 min or until voiding occurred. After three correct voids, time on toilet was decreased to 10 min. Nate often indicated when he needed to go to the toilet by taking his mother to the bathroom. Reinforcement intervals were given whether he had voided or not and were increased from 60, 75, 90 to 120 min when he stayed dry for three intervals. After an accident, Nate received a verbal reprimand and was taken to the toilet to see if he continued voiding.	0–1.3	0.5–0.3
Jonah–2	Day care centre	Increased liquid intake Response restriction Toy play on toilet Most to least prompting and praise Increasing the reinforcement interval Positive practice	150 ml fruit with soda in every reinforcement interval. Time between drinking and going to the toilet was prolonged from 0 to 30 min. Reinforcement intervals were increased from 10, 15, 20, 30, 45, 60, 75 up to 90 min after every third consecutive correct void, without accidents in the reinforcement interval. During reinforcement intervals he played in the hallway next to his group. During the 90 min interval, he was brought to his own group.	0.2–2	1.8–1.8

generalization phase, as the first training did not lead to a reduction in accidents (see Table II and Figure 1).

Recording

Dependent measures were correct voids and accidents. A correct void was defined as a urinary stream in the toilet. A urinary void was labeled as an accident when the void occurred elsewhere and when the void went through the participants' underwear (e.g. drops of urine in the underwear was not considered an accident). During baseline and training, correct voids and accidents were recorded, including date and time of the void. Trainers also noted related behaviors during training, for instance straining (i.e. raising intra-abdominal pressure to void), holding maneuvers (i.e. behaviors indicative of a compelling need to void) and facial expression while voiding.

Maintenance data were collected through a questionnaire that was completed by the primary caretaker. The mean daily or weekly frequency of toileting visits, number of correct voids, defecations and accidents, and nighttime incontinence were recorded. Follow-up data were collected after 18 months for Mandy, Daniel and Ella, after 6–9 months for Nate, Alice and Jonah, and after 3 months for Kevin. Parents and caretakers were also asked about their perspective on the training (e.g. if the training effect had maintained or diminished and which factors they considered to have contributed to the long-term outcome).

Reliability

Toilet training protocols were administered by trained master students. In 33% of training days (during baseline, training and generalization), a second observer (first author) was present to supervise the trainers and to record the number of correct voids and accidents. There was a 100% agreement between the trainer and the first author (correct voids and accidents).

Results

Figure 1 shows the daily frequency of correct voids and accidents during baseline, training and generalization phases. The mean frequency of correct voids and accidents during the baseline and generalization phase per participant can be found in Table II. Visual inspection shows that most children benefited from the toilet training. For Ella, Mandy, Nate, Alice and Kevin correct voids during the baseline phase did not occur with a single exception ($M = 0.05$ correct voids per day, $SD = 0.21$, range = 0–0.2), but higher number of correct voids occurred during the generalization phase ($M = 2.32$ correct voids per day, $SD = 1.25$, range = 1.3–3.6). In these children, apart from Ella, the number of accidents per day decreased (baseline: $M = 1.22$, $SD = 0.94$, range = 0.5–1.8; generalization: $M = 0.11$, $SD = 0.32$, range = 0–0.5). No difference in accidents between the baseline and generalization phase was seen in Ella as her only accident in the entire training occurred on the final day of the generalization phase.

In Daniel's case, correct voids were seen during baseline. Daniel was ill during the third day of training (data point 7), probably explaining the absence of correct voids and his relative high number of accidents on that day. During the

Table III. General outlines for the baseline and generalization phase.

Baseline phase	Participants stayed at their group and followed their own curriculum. Normal fluid intake. Participants were brought to the toilet every hour and were required to stay on the toilet for 5 min or until voiding occurred. Least-to-most prompting was used to promote correct sitting. When accidents occurred, the trainer neutrally expressed that the participant had voided in her/his pants and changed the pants.
Generalization phase	Participants stayed at their group and followed their own curriculum. Normal fluid intake. Participants were brought to the toilet every one-and-a-half to two hours, trials lasted for a maximum of 5–10 min or until voiding occurred. Prompts and rewards were faded out Similar consequences for accidents during the training and generalization phase (see Table IV). Training was transferred to caregivers, teachers and/or parents; they received the training outline on paper and were coached throughout the generalization process. In consultation with all persons involved, the diaper was removed outside training hours.

Table IV. Treatment components, their description, and their application.

Treatment component	Description	Application
Increased liquid intake	At the onset of the training phase, liquid intake was increased. When training progressed, liquid intake was brought back to normal levels with a 50/100 ml decrease alongside every increase of the reinforcement interval.	Increases the need to void and creates more learning opportunities.
Diaper removal	The diaper was removed during training hours in the three phases of the study. The participant walked around in normal, but light clothes to easily spot accidents.	To directly see and act upon accidents, and to make the participants aware of accidents, which might be perceived as aversive.
Response restriction	To block or restrict all behavior other than standing in front of the toilet, lowering pants, sitting down on the toilet and voiding in the toilet. Motor limitations, distractibility and hyperactivity made standing impossible; participants were allowed to sit on the toilet during toileting visits.	To stimulate correct voiding, as it is the only behavior that is not restricted.
Toy play on toilet seat	Some participants were given a toy to play with while sitting on the toilet seat to prevent them from playing with the water in the toilet bowl.	To prevent overexcitement by playing with the water and to focus on voiding.
Most-to-least prompting strategy and praise	Participants were given verbal, model and physical prompts to stay seated on the toilet seat and to void in the toilet. Prompts were given approximately every 5 min. After correct voiding, participants were given a reinforcement interval in which they received praise and tangibles. Both were faded out when the number of correct voids increased.	To promote and reinforce correct sitting on toilet seat and voiding.
Reinforcement interval in the natural environment	Following a correct void, the participant was rewarded and allowed to go back to his or her normal curriculum for a certain amount of time, depending on the training-progress. This period is called the ''reinforcement interval''.	To normalize the day, to foster generalization of training results and to prevent stress and frustration on part of the participant.
Increasing the reinforcement interval	The reinforcement interval was increased when the participant had stayed dry for two or three reinforcement intervals.	
Decrease in time-on-toilet	When a decrease in time-on-toilet before voiding was seen (usually after two or three voids in the training phase), toileting visits were ended after a correct void or after 10 min, unless clear signs of straining were seen. Irrespective of voiding, the participant was given a reinforcement interval, but only received praise and tangibles when voiding had occurred.	To prevent the undermining of the developed association of the toilet seat and voiding.
Similar consequences for accidents during the training and generalization phase	The participants received the same consequences for accidents during both training and generalization phase.	Little accidents occurred throughout the study and with this intervention, learning opportunities were increased.
Remain seated on toilet following an accident	Participants had to stay on the toilet until they voided or clearly tried to void, which was followed by a reinforcement interval.	To strengthen the association between urinating in the toilet and reinforcement interval. Was used when caretakers and/or trainers expected or saw that positive practice was too frustrating for the participant, as this would counter training compliance.
Positive practice	Participants were prompted to go to the toilet, lower their pants, sit on the toilet for 1–3 s and pull up their pants. This sequence was repeated three times in different locations.	To train the entire toileting sequence and prevent more accidents. Was only used when application was not too frustrating (see ''Remain seated on toilet following an accident'').

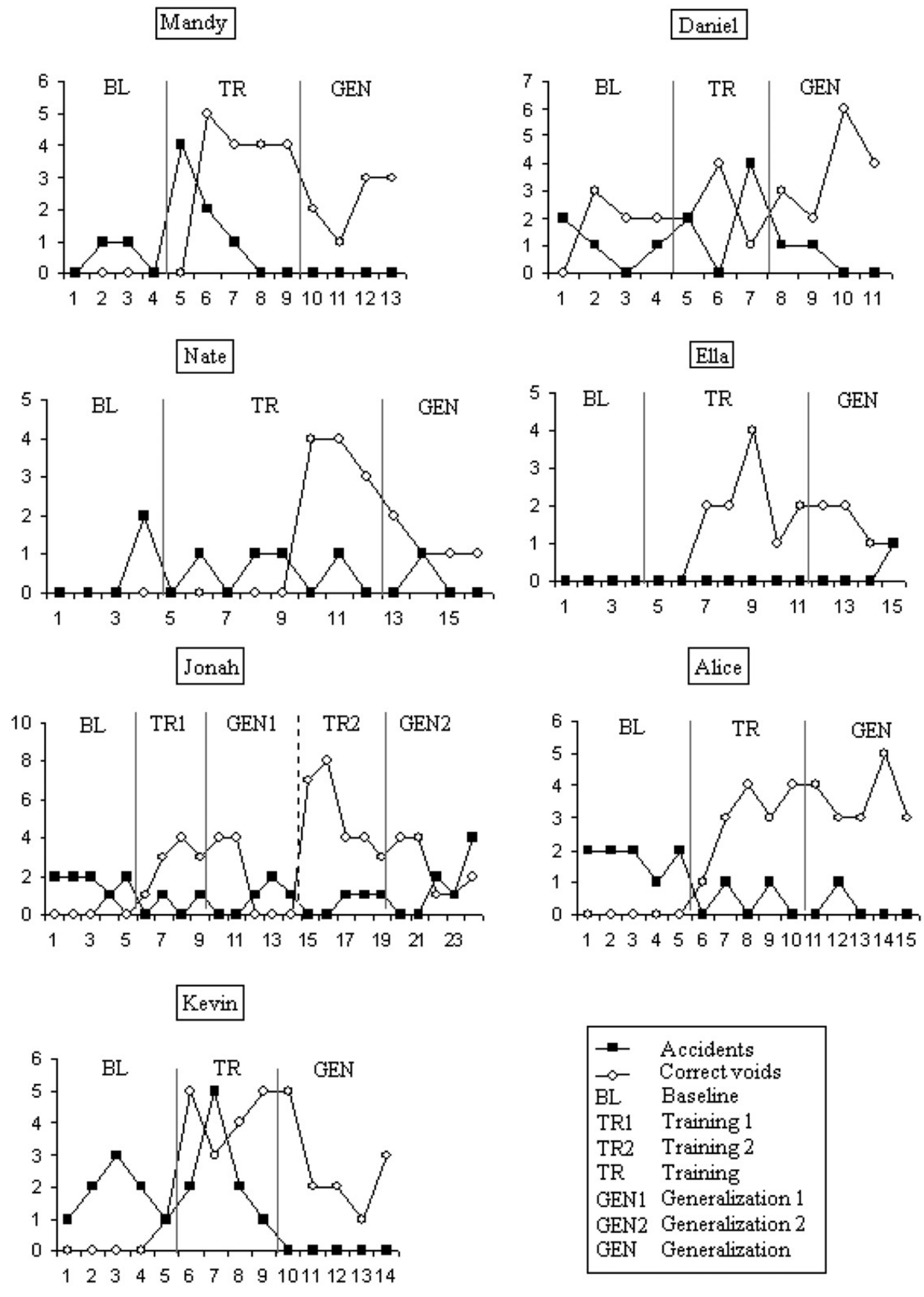

Figure 1. Frequency of correct voids and accidents per participant.

generalization phase, a clear rise in correct voids and a decline in accidents were evident. Training for Jonah was only partially successful; his correct voids increased during both generalization phases compared to baseline (0.2 vs. 1.6–1.8 correct voids per day), but his number of accidents did not change (1.6 vs. 0.8–1.8 accidents per day).

Changes in the training protocols with four of the participants: Mandy (data point 6), Ella (data point 7), Nate (data point 11) and Jonah (initiation of TR2), were associated with increases in correct voids. At the start of these training days, Mandy and Jonah were required to remain seated until voiding occurred instead of leaving the toilet after 10 min and Jonah also received a further increase in fluid intake. In Nate and Ella, training was transferred from their day care center to their homes at this point in the intervention and, in Nate's case; his mother was introduced as the trainer.

The mean duration of the entire training protocol in days across all participants was 15.4 d ($SD = 4.1$, range 11–24 d).

Follow-up

Information gathered at follow-up revealed that Ella, Alice and Nate still voided on the toilet three to five times per day and urinary accidents did not occur. Response generalization was reported in Ella; her primary caretakers indicated that she became continent for feces during and following the toilet training and nighttime incontinence decreased. When her toileting visits occurred relatively late at night and early in the morning, she often stayed dry at night. Kevin showed self-initiated toileting during the day and could, according to his parents, appropriately answer questions about his needing to go to the toilet or not. Training results for Mandy, Daniel, Nate and Jonah were not maintained; they wore diapers during the day and of these four only Jonah occasionally voided or defecated on the toilet.

Conclusions

In this study, modified RR training protocols were used to toilet train seven individuals with AS. Along with increases in correct voids and decreases in accidents, the reinforcement interval was prolonged, time-on-toilet was decreased and the liquid intake was returned to normal quantities. Individuals were trained in their natural environment and received prompts and consequences for correct voids and accidents to encourage correct voiding.

With all participants, correct voids were seen more frequently during the generalization phase as compared to the baseline phase. In four participants, a decrease in accidents between the baseline and generalization phase occurred, in association with the training. In the other three participants no differences in accidents were seen between both phases; accidents remained low (≤ 2) in Ella and Nate and high (≥ 6) in Jonah. Follow-up questionnaires indicate that positive training results were maintained in three participants. In addition, in Ella continence had generalized to feces and nighttime continence.

Although the caregivers primarily attributed the observed relapses with Mandy, Daniel, Nate and Jonah to environmental factors such as too many changes in routines in daycare facilities, training factors could also have contributed to the relapses, as Didden et al. [8] reported successful maintenance of training effects over time while using a modified Azrin–Foxx procedure. Differences in long term outcome between our and Didden et al. their study could be a function of differences between the two protocols, components present in their training, that were absent in ours (e.g. rewards for dry pants, restitutional overcorrection, 20 min time-on-toilet during scheduled toileting visit) or vice versa (e.g. positive practice, decrease of fluid intake during training). Alternatively, differences in long-term training outcomes could be a function of the more extended post training phases included in Didden et al. their study during which participants were monitored and trained during waking hours for almost two months after the initial training phase with the training elements being faded more slowly. Overall, these differences may have resulted in more training opportunities for the participants in Didden et al. their study, possibly leading to a stronger discrimination between a correct void and an accident. Further research seeking to determine the additional benefits of each training component is clearly justified. Finally, our findings stress the need for continuing efforts and patience from the caretakers in all living and daytime environments to promote maintenance of continence [11].

Other additional training components could also have been implemented during the training phase to foster maintenance. One of these components is a communication-mode with which children can indicate their need to void. Although the goal of the training was prompted voiding, two participants (Nate and Kevin) showed self-initiated toileting during the training and generalization phase. They had started to imitate a gesture for toileting made by the trainer. Future research investigating the effect of communication training as an element of toilet training in AS is warranted. Second, as fecal incontinence is a risk factor for urinary incontinence [6] fecal continence should be trained alongside or following training for urinary continence. In the training and generalization phases, several participants defecated in the toilet but fecal accidents occurred as well, indicating that additional training for fecal continence is required (see e.g. [12])

Besides these training components, medical factors should be taken into consideration. Lower baseline frequencies of accidents were found in individuals with AS when compared to frequencies of individuals with severe ID [9, 11]. This may be indicative of an underactive bladder [13]. An underactive bladder is often accompanied by straining and an interrupted urinary flow pattern, which were also seen in the participants in our study. Further, during the training phase holding maneuvers were seen only once despite the increased liquid intake (up to 2000–3000 ml). This absence of holding maneuvers is a remarkable finding since most accidents consisted of large quantities of urine which could not be interrupted by the trainers, suggesting that the bladder was full enough to sense an urge to void. Future research should focus on abnormal voiding patterns and characteristics in individuals with AS.

Our results should be interpreted in the light of the following limitations. Because of the low number of participants and individualized training protocols, no firm statements can be made about the generalization of the results and about which training components were responsible for the individual results. Another limitation is the use of AB designs. Due to practical constraints and time limitations we did not use a control group or multiple baseline design.

Despite obvious limitations, our study suggests that urinary continence is a feasible goal in the lives of individuals with AS. Further research is needed to evaluate procedures with which initial treatment effectiveness can be maintained long-term in natural environments. Also, qualitative (low voiding frequencies) and quantitative (e.g. straining, absence of holding maneuvers) voiding characteristics observed during training, suggest that individuals with AS may have voiding dysfunctions. Future studies should explore this, as this could impact toilet training protocols and medical treatments.

Acknowledgments

The authors would like to thank participants, parents and caretakers for participating in this research. Gratitude goes out to Louke Peters, Mieke van der Veen and Elles Volker for their assistance during toilet training. Finally, the authors wish to thank Marije Averink and Eef Rasing for their advice.

Declaration of interest

This research was funded by the Angelman Foundation, The Netherlands. The authors report no conflicts of interests. The authors alone are solely responsible for the content and writing of this paper.

References

1. Lalande M, Calciano MA. Molecular epigenetics of Angelman syndrome. Cellular and Molecular Life Sciences 2007;65:947–960.
2. Williams CA. The behavioral phenotype of the Angelman syndrome. American Journal of Medical Genetics 2010;154C: 432–437.
3. Buntinx IM, Hennekam RCM, Brouwer OF, Stroink H, Beuten J, Mangelschots K, Fryns JP. Clinical profile of Angelman syndrome at different ages. American Journal of Medical Genetics 1995;56: 176–183.
4. Laan LAEM, den Boer ATh, Hennekam RCM, Renier WO, Brouwer OF. Angelman syndrome in adulthood. American Journal of Medical Genetics 1996;66:356–360.
5. Cicero FR, Pfadt A. Investigation of a reinforcement-based toilet training procedure for children with autism. Research in Developmental Disabilities 2002;23:319–331.
6. Kroeger KA, Sorensen-Burnworth R. Toilet training individuals with autism and other developmental disabilities. Research in Autism Spectrum Disorders 2009;3:607–618.
7. Landefeld CS, Bowers BJ, Feld AD, Hartmann KE, Hoffman E, Ingber MJ, King Jr JT, McDougal S, Nelson H, Orav EJ, et al. National institutes of health state-of-the-science conference statement: Prevention of fecal and urinary incontinence in adults. Annals of Internal Medicine 2008;148:449–460.
8. Didden R, Sikkema PE, Bosman ITM, Duker PC. Use of a modified Azrin–Foxx toilet training procedure with individuals with Angelman syndrome. Journal of Applied Research in Intellectual Disabilities 2001;14:64–70.
9. Duker PC, Averink M, Melein L. Response restriction as a method to establish diurnal bladder control. American Journal on Mental Retardation 2001;3:209–215.
10. Sigafoos J, Didden R, O'Reilly M. Effects of speech output on maintenance of requesting and frequency of vocalizations in three children with developmental disabilities. Augmentative and Alternative Communication 2003;19:37–47.
11. Averink M, Melein L, Duker PC. Establishing diurnal bladder control with the response restriction method: Extended study on its effectiveness. Research in Developmental Disabilities 2005;26: 143–151.
12. Matson JL, LoVullo SV. Encopresis, soiling and constipation in children and adults with developmental disability. Research in Developmental Disabilities 2009;30:799–807.
13. Nevéus T, von Gontard A, Hoebeke P, Hjälmås K, Bauer S, Bower W, Jørgensen TM, Rittig S, Van de Walle J, Yeung CK, et al. The standardization of terminology of lower urinary tract function in children and adolescents: Report from the standardization committee of the international children's continence society (ICCS). Neurourology and Urodynamics 2007;26: 90–102.
14. De Bildt AA, Kraijer DW. Vineland-Z: Sociale zelfredzaamheidschaal voor kinderen en jeugdigen met een verstandelijke beperking. [Vineland Adaptive Behavioral Scales, Survey Form]. Leiden, The Netherlands: PITS; 2002.
15. Van der Meulen BF, Ruiter SAJ, Lutje Spelberg HC, Smrkovsky M. Bayley Scales of Infant Development – Second Edition – Dutch Version (BSID-II-NL). Lisse, the Netherlands: Swets & Zeitlinger; 2002.