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Incontinence in individuals with Angelman syndrome: A comparative study

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ABSTRACT

Frequency and type of incontinence and variables associated with incontinence were assessed in individuals with Angelman syndrome (AS; $n = 71$) and in a matched control group ($n = 69$) consisting of individuals with non-specific intellectual disability (ID). A Dutch version of the "Parental Questionnaire: Enuresis/Urinary Incontinence" (Beetz, von Gontard, & Lettgen, 1994) was administered and information on primary caretakers' perspectives regarding each individual's incontinence was gathered. Results show that diurnal incontinence and fecal incontinence during the day more frequently occurred in the control group than in the AS group. In both groups, nocturnal enuresis was the most common form of incontinence. More incontinence was seen in individuals with AS who were younger, had a lower level of adaptive functioning and/or had epilepsy. Individuals with AS were able to stay dry for longer periods of time than the controls and often showed both in-toilet urination and urinary accidents during the day, whereas accidents and correct voids during the day were more set apart in the control group. Also, persons with AS had a lower micturition frequency implying possible voiding postponement. Both groups showed high rates of LUTS (lower urinary tract symptoms) possibly indicative of functional bladder disorders such as voiding postponement, dysfunctional voiding, or even an underactive bladder. In general, most primary caretakers reported severe intellectual disability as the main cause for urinary incontinence. Based on these results incontinence does not appear to be part of the behavioral phenotype of Angelman syndrome. Therefore, pediatric or urologic diagnostics and treatment are recommended for all persons with incontinence and intellectual disability. Further implications for practice and research are given.

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1. Introduction

Angelman syndrome (AS) is a neurodevelopmental disorder caused by the absence or malfunctioning of expression of maternally imprinted genes in the region at 15q11-13 (Lalande & Calciano, 2007). Individuals with AS often suffer from epilepsy and their behavioral phenotype includes severe intellectual disability (ID), motor and speech deficits, easy evocable and sometimes inappropriate laughter, and sleep disturbances (Clayton-Smith & Laan, 2003; Didden, Korzilius, Smits, & Curfs, 2004; Williams, 2010). The developmental age of individuals with AS rarely exceeds two years (Peters et al., 2004) suggesting that development of continence may be severely delayed, as continence in typically developing children is often achieved at the age of three years (Schum, Kolb, McAuliff, Simms, & Underhill, 2002).

Incontinence can be defined as uncontrollable leakage of urine in individuals older than five years, for 2 or more episodes per week (DSM-IV; American Psychiatric Association, 2000). It can be further classified as continuous and intermittent incontinence during the day (diurnal incontinence; DI) and/or during sleep (enuresis of nocturnal enuresis; NE). DI comprised a heterogeneous groups of disorders, including overactive bladder, voiding postponement, dysfunctional voiding and underactive bladder. When an individual is continent, but relapses for a period of six months or longer, this is called secondary incontinence, if the longest interval is less than 6 months, incontinence is termed primary. If lower urinary tract symptoms (LUTS) are present, the term non-monosymptomatic NE is chosen – without LUTS it is termed monosymptomatic NE. Fecal incontinence (FI) is defined as voluntary and involuntary defecation in inappropriate places occurring at least once per month for three consecutive months (according to DSM-IV). FI can occur with functional constipation or as non-retentive fecal incontinence.

Incontinence may result in social stigmatization, anxiety, physical discomfort, and urinary tract infections, which may lead to dependency of caregivers, exclusion from peer groups and certain activities (Cicero & Pfadt, 2002; Kroeger & Sorensen-Burnworth, 2009; Mehta et al., 2003). In addition to adverse effects for the individuals themselves, incontinence can place a burden on the lives of family members (Gotoh et al., 2009) and costs for diapers and medical treatments are often considerable (Borrie & Davidson, 1992; Landefeld et al., 2008).

Prevalence studies of incontinence in AS are scarce and have yielded inconsistent outcomes. For example, Buntinx et al. (1995) showed that 14 (63%) of the children and youngsters aged 2–16 years with AS suffered from DI, whereas two (12.5%) of the individuals with AS aged 16 and older were incontinent. A study on adults with AS aged 20–53 years ($N = 28$) showed that 43% of the individuals were incontinent when they were sent to the toilet at regular scheduled times during the day (Laan, den Boer, Hennekam, Renier, & Brouwer, 1996). Didden et al. (2004) found that 93% out 133 of individuals with AS between five and 44 years old suffered from NE and FI; however, DI was not studied.

The previous research on incontinence in people with AS has several limitations. Specifically, none of the studies included a control group, nor did they address all types of incontinence, and sample sizes were relatively small. Further, detailed information about incontinence was not given and relationships between incontinence and associated variables were not explored. The aim of this current study is to gain more insight regarding the nature and prevalence of incontinence in AS by increasing the sample size, including all types of incontinence, and comparisons with a control group. Further, the inclusion of a matched control group allows this study to test the hypothesis that incontinence may be part of the behavioral phenotype of AS (Dykens, 1995). Finally, the present study extends our knowledge on AS as it also explores associations between incontinence and other variables in individuals with AS.

2. Materials and methods

2.1. Participants

Participants in the AS group ($N = 71$; 36 male) had a mean age of 20.5 years ($SD = 9.8$; range 6.8–45.8), a mean developmental age of 1.4 years ($SD = 0.10$; range 0.4–2.9) and their motor abilities reached a mean age of 1.8 years ($SD = 0.9$; range 0.4–3.2). Most participants had the deletion subtype ($n = 45$, 63%), followed by mutation ($n = 6$, 9%) and uniparental disomy ($n = 5$, 7%); in the remaining participants the cause of AS was unknown or not reported ($n = 15$, 21%). The majority ($n = 59$, 83%) of participants also suffered from epilepsy of whom 43 (61%) successfully used anticonvulsive medication to control seizures. Eleven (16%) participants were wheelchair bound and 24 (36%) participants lived in a facility.

Participants in the control group ($N = 69$; 37 male) had a mean age of 22.9 years ($SD = 12.6$; range 5.3–48.11), a mean developmental age of 1.3 years ($SD = 1.2$, range 0.2–3.7) and their motor abilities reached a mean age of 1.3 years ($SD = 1.3$, range 0.0–4.5). In 21 individuals (34%) the cause of the ID was known, 13 causes were mentioned including Down syndrome ($n = 3$) and oxygen deficiency at birth ($n = 2$). The larger part ($n = 44$, 64%) of participants suffered from epilepsy of whom 10 (27%) successfully used anticonvulsive medication to control seizures. Thirty-three (50%) participants were wheelchair bound and 42 (68%) participants lived in a facility.

Individuals' characteristics were compared between the two groups. Differences in gender distribution ($\chi^2(1) = 0.12$, $p = .73$), age ($t(128.72) = 1.21$, $p = .23$), level of adaptive functioning ($t(114.15) = 0.58$, $p = .56$), motor abilities ($t(107.81) = 1.70$, $p = .09$), and presence of urinary tract infections ($\chi^2(1) = 0.01$, $p = .95$) were not statistically significant. However, more participants from the CG lived in a facility ($\chi^2(1) = 12.07$, $p < .01$) and were wheelchair bound ($\chi^2(1) = 18.68$, $p < .01$). Fewer participants suffered from epilepsy ($\chi^2(1) = 6.73$, $p = .01$) as compared to the AS group.