Research in Developmental Disabilities xxx (2016) xxx–xxx

Contents lists available at ScienceDirect

Research in Developmental Disabilities

Incontinence and psychological symptoms in individuals with Mowat-Wilson Syndrome

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ARTICLE INFO

Article history:
Received 21 July 2016
Received in revised form 3 November 2016
Accepted 4 January 2017
Available online xxx
Number of reviews completed is 2

Keywords:
Mowat-Wilson Syndrome
Urinary incontinence
Fecal incontinence
Enuresis
Psychopathology

ABSTRACT

Background: Mowat-Wilson Syndrome (MWS) is caused by deletion/mutation of the *ZEB2* gene on chromosome 2q22. MWS is characterized by a distinctive facial appearance, severe intellectual disability and other anomalies, e.g. seizures and/or Hirschsprung disease (HSCR). Most individuals have a sociable demeanor, but one third show psychological problems.
Aims: The aim was to investigate incontinence and psychological problems in MWS.
Methods and procedures: 26 children (4–12 years), 13 teens (13–17 years) and 8 adults (>18 years) were recruited through a MWS support group. The Parental Questionnaire: Enuresis/Urinary Incontinence, as well as the Developmental Behaviour Checklist (DBC) were completed by parents or care-givers.
Outcomes and results: 97.7% of persons with MWS had incontinence (nocturnal enuresis 74.4%; daytime urinary incontinence 76.2%; fecal incontinence 81.4%). Incontinence remained high over age groups (children 95.8%, teens 100%, adults 100%). 46.2% of children, 25% of teens and 37.5% of adults exceeded the clinical cut-off on the DBC. The ability to use the toilet for micturition improved with age.
Conclusions and implications: MWS incontinence rates are very high. All had physical disabilities including anomalies of the genitourinary and gastrointestinal tract. Due to the high prevalence rates, a screening for incontinence and psychological problems in MWS is recommended.

What this paper adds?

This study is the first assessment of incontinence in a group of children, teens and adults with MWS. Results show very high prevalence of incontinence, with no significant differences between age groups. Although incontinence rates remain high, some adaptive toileting skills are present in adults with MWS. Due to physical disabilities both, functional and organic incontinence could be present.

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http://dx.doi.org/10.1016/j.ridd.2017.01.006
0891-4222/

1. Introduction

1.1. Mowat-Wilson Syndrome

Mowat-Wilson Syndrome (MWS) is a congenital syndrome caused by a deletion or mutation of the *ZEB2* gene on chromosome 2q22 (Mowat & Wilson, 2010, Chapter 35). It was first described by Mowat et al. (1998) in six individuals with symptoms of severe intellectual disability (ID), microcephaly, short stature and Hirschsprung disease (HSCR) (Mowat et al., 1998). Further characteristics of MWS are typical facial features (prominent nasal tip and chin, cupped ears with upturned lobes, deep-set eyes, hypertelorism, broad medially sparse eyebrows), severe developmental delay, seizures, constipation and a happy, social demeanor (Adam et al., 2006; Mowat & Wilson, 2010, Chapter 35). Prevalence is estimated at 1: 50,000–70,000 (Mowat & Wilson, 2010, Chapter 35).

1.2. Behavioral phenotype of Mowat-Wilson Syndrome

The behavioral phenotype in MWS was examined by Evans et al. in a sample on 61 individuals, who found less depressive or mood problems, more oral (as chewing objects or grinding teeth) and stereotyped behaviours and an underreaction to pain in comparison to a group of non-syndromatic persons with ID (Evans et al., 2012). Further descriptions of MWS point out the happy demeanor, sociability and frequent laughter, but also self-injuring and hyperactive behavior (Adam, Justice, Bean, & Fernhoff, 2008; Mowat & Wilson, 2010, Chapter 35). Sleep disturbances with an association to psychological problems are also found in MWS (Evans, Mowat, Wilson, & Einfeld, 2016). Intelligence in MWS is mostly in the range of severe ID (Evans et al., 2012; Garavelli et al., 2009).

1.3. Incontinence

Functional incontinence (including nocturnal enuresis (NE), daytime urinary incontinence (DUI) or fecal incontinence (FI)) is a common disorder affecting about 1–10% of typically developing children and decreasing to <1% in adolescence and adulthood (Franco, Austin, Bauer, von Gontard, & Homsy, 2015). In individuals with ID, incontinence rates are much higher (approx. 30–40%), and are associated with the level of ID (von Wendt, Simila, Niskanen, & Jarvelin, 1990). In a population-based sample, 33–38% of 7-year-old children with severe ID had some subtype of incontinence, and at the age of 20 years, 19–24% of individuals with severe ID still had incontinence problems (von Wendt et al., 1990).

1.4. Incontinence in Mowat-Wilson Syndrome

Incontinence has not been systematically examined in MWS. In a report on 12 cases with MWS aged 0–23 years, 3 of the 8 individuals who were older than 4 years were not toilet trained, one was in the process of toilet training and 3 were continent (Adam et al., 2006). HSCR is a condition often associated with constipation and FI and is found in approximately 45% of MWS patients (Coyle & Puri, 2015). In a review of published cases, constipation was reported in 30%, genitourinary and renal anomalies in 52% (Garavelli et al., 2009).

1.5. Aim of the study

The aim of this study was to examine incontinence subtypes, associated conditions (constipation, lower urinary tract symptoms (LUTS), anomalies of the genitourinary and gastrointestinal tract), adaptive toileting skills and behavioral symptoms in different age groups of individuals with MWS.

2. Material and methods

2.1. Procedure

Packages with questionnaires were sent to all member families of the German Mowat-Wilson Syndrome support group "Mowat-Wilson Deutschland". As it comprises only 10 families, the study was also announced on the homepage of the International Mowat-Wilson Foundation. Questionnaires in four languages were provided (English, German, French, Italian), which could be requested online by interested families. Following informed consent, parents or caregivers were asked to complete the questionnaires and send them back. Questionnaires were sent out and received over 28 months (June 2013–October 2015).

2.2. Instruments

Incontinence was assessed by the "Parental Questionnaire: Enuresis/Urinary Incontinence" (von Gontard, 2012b), "Encopresis Questionnaire – Screening Version" (von Gontard, 2012a). Six questions about adaptive toileting skills were added

Please cite this article in press as: Niemczyk, J., et al. Incontinence and psychological symptoms in individuals with Mowat-Wilson Syndrome. *Research in Developmental Disabilities* (2016), http://dx.doi.org/10.1016/j.ridd.2017.01.006