



# Incontinence in persons with Noonan Syndrome

Justine Niemczyk<sup>a</sup>, Monika Equit<sup>a</sup>, Sorina Borggreffe-Moussavian<sup>a</sup>, Leopold Curfs<sup>b</sup>, Alexander von Gontard<sup>a</sup>

<sup>a</sup>Department of Child and Adolescent Psychiatry, Saarland University Hospital, Homburg, Germany

<sup>b</sup>Department of Clinical Genetics, Governor Kremers Centre, Maastricht University Medical Centre, Maastricht, The Netherlands

Correspondence to:  
J. Niemczyk, Department of Child and Adolescent Psychiatry, Saarland University Hospital, 66421 Homburg, Germany. Tel.: +49 6841 16 24385, fax: +49 6841 16 24397

Justine.niemczyk@uks.eu (J. Niemczyk)  
Monika.equit@uks.eu (M. Equit)  
info@frische-koepfe.de (S. Borggreffe-Moussavian)  
leopold.curfs@maastrichtuniversity.nl (L. Curfs)  
Alexander.gontard@uks.eu (A. von Gontard)

## Keywords

Noonan Syndrome; Functional incontinence; Enuresis; Fecal incontinence; Daytime urinary incontinence; Psychological symptoms

Received 20 November 2014  
Available online 18 June 2015

## Summary

### Objective

Noonan Syndrome (NS) is an autosomal neurodevelopmental disorder with a high phenotypic variability. Mutations in several genes of the RAS-MAPK signaling pathways are now known to be responsible for NS. Most of the children with NS are of average intelligence, one-third have a mild intellectual disability (ID) (IQ 50–79). So far, no studies have assessed incontinence in persons with NS. The aim of this study therefore was to investigate the prevalence of incontinence and psychological problems in persons with NS.

### Subjects and methods

Nineteen children (5–17 years) and 10 adults (18–48 years) with NS were recruited through a German parent support group (58.6% male, mean age 15.26 years). The “Parental Questionnaire: Enuresis/Urinary Incontinence”, “Encopresis Questionnaire – Screening Version” and the German version of the International Consultation on Incontinence Questionnaire – Pediatric Lower Urinary Tract Symptom (ICIQ-CLUTS) were completed by parents or caregivers to assess incontinence and lower urinary tract symptoms (LUTS). The Developmental Behavior Checklist for parents (DBC-P) or the Developmental Behavior Checklist for adults (DBC-A) were filled out to assess psychological symptoms.

## Results

In total, 27.3% of the children (4–12 years) had nocturnal enuresis (NE), 36.4% had daytime urinary incontinence (DUI), and 11.1% had fecal incontinence (FI). Only one adolescent (13–17 years) had NE (14.3%) and one young adult (18–30 years) had FI (11.1%); 36.4% of the children, 33.3% of the adolescents and 12.5% of young adults had a DBC score in the clinical range. No adult (>30 years) had incontinence or a critical DBC score.

Children and adolescents with NE had significantly higher scores in the DBC total score as well in the “self-absorbed” and “social relating” subscales than continent children and adolescents, whereas no significant difference was found between children and adolescents with DUI compared with the continent group.

## Conclusions

A significant proportion of children with NS are affected by incontinence. Incontinence is a relevant problem in children and adolescents with NS, but does not persist into adulthood. In particular, psychological problems are present in children and adolescents with NE. Screening for both incontinence and psychological symptoms are recommended in children with NS. As most of the children with NS have average intelligence or a mild ID, they can be treated effectively with standard methods.

Table

	Total sample <i>n</i> = 29
Male <i>n</i> (%)	17 (58.6)
Mean age in years (SD)	15.27 (8.6)
At least one subtype of incontinence <i>n</i> (%)	6 (26.1)
NE <i>n</i> (%)	4 (16.0)
DUI <i>n</i> (%)	4 (13.8)
FI <i>n</i> (%)	2 (7.4)
DBC-P/A Total score >clinical cut-off <sup>a</sup> <i>n</i> (%)	6 (35.3)/1 (10)

<sup>a</sup> Clinical Cut-off for the DBC-P is a Total score >46, clinical Cut-off for the DBC-A is a Total score >51 [21].

## Introduction

Noonan Syndrome (NS) is an autosomal dominant, neurodevelopmental disorder with a high phenotypic variability. It was first described in 1963 by Noonan and Ehmke [1] in a group of patients with similar anomalies. The prevalence of NS is 1 in 1,000–2,500 births [2]. As shared genetic cause, mutations in several genes of the RAS-MAPK signaling pathways on the chromosomal band 12q24.1 were found [2]. Recent overviews show that about 50% of all persons with NS have a mutation in the PTPN11 gene, 10% in the SOS1 gene, 10% in RAF1, <2% in KRAS, and <1% in NRAS. Other genes affected are BRAF and SHOC2 [2,3].

Typical signs and symptoms in NS are congenital heart defects, auditory deficits, short stature, abnormal pigmentation, cryptorchidism, and typical facial features (ptosis, hypertelorism, low-set ears, depressed nasal root) [2,4]. About 20–30% have an intellectual disability (ID) (IQ < 70), a further 37% have a below average or borderline (70–89) IQ [5].

There are only few publications on psychological problems in NS. Behavioral and developmental difficulties were described in about 25–50% of children with NS [5–7]. The most common symptoms reported in children with NS were stubbornness, clumsiness, mood and communication problems, and faddy eating [7]. In adults with NS, disorders such as panic disorder, alexithymia, obsessive–compulsive disorder, bipolar disorder, schizophrenia, anxiety, and depression can occur [8,9]. Higher levels of alexithymia (inability of expressing emotions) and social distress were found in 40 adults with NS compared with a control group [9].

Functional incontinence such as nocturnal enuresis (NE), daytime urinary incontinence (DUI), and fecal incontinence (FI) are common in normally developed children, with prevalence rates of 10% for NE, 2–3% for DUI, and 1–3% for FI in 7-year-old children [10]. In children with special needs, for example children with ID, incontinence rates are much higher [11]. Also, children with incontinence are more often affected by psychological disorders; the rates are especially high in children with FI [12–14].

The aim of this study was to investigate the prevalences of incontinence, lower urinary tract symptoms (LUTS), and psychological problems in persons with NS. It was hypothesized that incontinence occurs more often in NS than in the normal population. Another aim was to analyze the associations between psychological problems and incontinence in persons with NS.

## Material and methods

Subjects were recruited through a German Noonan syndrome support group. Packages with questionnaires, information sheets, and return envelopes for children and adults were sent to the chairperson of the support group, who distributed them to all 107 member families. Following informed consent, parents/caregivers were asked to complete the questionnaires and send them back anonymously. Twenty-nine questionnaires were returned. The study was approved by the local ethics committee.

Incontinence and LUTS were assessed by the “Parental Questionnaire: Enuresis/Urinary Incontinence” [15], “Encopresis Questionnaire – Screening Version” [16], and the German version of the International Consultation on Incontinence Questionnaire – Pediatric Lower Urinary Tract Symptom (ICIQ-CLUTS) [17]. NE and DUI were diagnosed from the age of 5 years when wetting occurs at least once per month, according to the International Children’s Continence Society (ICCS) criteria [18]. FI was diagnosed from the age of 4 years according to DSM-5 criteria, that is when soiling occurs at least once per month [19]. The 10 questions of the ICIQ-CLUTS build a LUTS score with clinically relevant scores higher than 13.

Behavioral and psychological symptoms were assessed with the German version of the Developmental Behavior Checklist (DBC) [20]. The parental questionnaire consists of 96 items, which are rated with “0, not true as far as you know”; “1, somewhat or sometimes true”; and “2, very true or often true”. Parents/caregivers of children and adolescents (<18 years) completed the parental version (DBC-P), those of adults (≥18 years) the adult version (DBC-A). The DBC-P comprises the subscales “Disruptive/Antisocial” (DA), “Self-Absorbed” (SA), “Communication Disturbance” (CD), “Anxiety” (A), and “Social Relating” (SR), and the “Total Behavior Problem Score” (TBPS). The DBC-A has the same subscales except that the fourth scale is termed “Anxiety/Depression” (AD). A TBPS above 46 in the DBC-P and above 51 in the DBC-A are considered as clinically relevant [21]. German norms are available for children and adults with mild, moderate, and severe intellectual disability for both the DBC-P and the DBC-A [21,22]. DBC-P/A Total values are expressed in T values, which belong to a standard norm scale with the mean = 50 and the standard deviation = 10. The DBC-P and DBC-A questionnaires were evaluated according to the norms of mild intellectual disability.

Completed questionnaires of 19 children (5–17 years of age, mean age = 11.2 years (SD = 4.1), 57.9% male), and 10 adults (18–48 years of age, mean age = 23.9 years (SD = 9.4), 60% males) were included in the study.

The data were analyzed with IBM SPSS Statistics 22, using descriptive statistics and performing nonparametric (chi-square tests, the Fisher exact test, Mann–Whitney U tests) for categorical data and parametric tests for interval data (Student *t* test, Welch test). Results were considered significant at  $p < 0.05$ .

## Results

The sample included 11 children between 4 and 12 years of age (39.3%), seven adolescents between 13 and 18 years of age (25.0%), nine young adults (18–30 years) (32.1%), and one adult older than 30 years (3.6%). In one case, age was not stated. Descriptive data and prevalence of incontinence in the four age groups are outlined in Table 1.

Of the total sample, 26.1% were affected by at least one subtype of incontinence. Within the subgroups, incontinence was higher in children than in adolescent and adult groups (44.4% vs. 14.3% vs. 16.7% vs. 0%). NE was found in 27.3% of children, 14.3% of adolescents, and in none of (young) adults. DUI was only present in children (36.4%). FI was found in one child and one young adult. No subject

Download English Version:

<https://daneshyari.com/en/article/4162184>

Download Persian Version:

<https://daneshyari.com/article/4162184>

[Daneshyari.com](https://daneshyari.com)