Autism Spectrum Disorders in Children with Functional Defecation Disorders

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Objective To prospectively assess the prevalence of autism spectrum disorder (ASD) symptoms in children presenting with functional defecation disorders.

Study design Children (age 4-12 years) with functional constipation or functional non-retentive fecal incontinence according to the Rome III criteria referred to a specialized outpatient clinic were included. Parents completed 2 validated ASD screening questionnaires about their child; the Social Responsiveness Scale (SRS) and the Social Communication Questionnaire-Lifetime (SCQ-L). A total SRS score of ≥51 is a strong indicator for the presence of ASD. On the SCQ-L, a score of ≥15 is suggestive for ASD.

Results In total, 242 patients (130 males, median age 7.9 years) were included. Of these, 91% were diagnosed with functional constipation and 9% with functional non-retentive fecal incontinence. Thirteen children (5.4%) had previously been diagnosed with ASD. Twenty-six children (11%) had both SRS and SCQ-L scores at or above cutoff points, strongly suggestive for the presence of ASD. Solely high SRS were present in 42 children (17%), whereas two children (1%) only had high SCQ-L scores. Altogether, 29% had ASD symptoms, indicated by SRS and/or SCQ-L scores at or above the cutoff values. These children were older than children without ASD symptoms and presented with a longer duration of symptoms.

Conclusions A substantial number of children (29%) presenting with a functional defecation disorder at a tertiary hospital has concomitant ASD symptoms. Clinicians should be aware of ASD symptoms in children with functional defecation disorders. *(J Pediatr 2013;163:873-8)*.

Functional defecation disorders, functional constipation (FC), and functional non-retentive fecal incontinence (FNRFI) are common problems in children. The world-wide prevalence of FC in children ranges from 0.7% to 29.6%.[1] In some children, fecal incontinence (FI) may occur without signs of fecal retention better known as FNRFI.[2] FNRFI has a reported prevalence in children of 1.5% to 9.8%.[3] Despite maximal appliance of current conservative treatment modalities, long-term follow-up data show that functional defecation disorder symptoms persist into adulthood in 25%-30% of children, negatively affecting quality of life.[4] The pathophysiology of functional defecation disorders is multifactorial and largely unknown.[5,6] A high prevalence of gastrointestinal disorders, most frequently constipation, has been reported in children with autism spectrum disorders (ASD).[7-12] Individuals affected by ASD may share a common triad of impairment in social interactions, impaired and atypical verbal and non-verbal communication, and repetitive and unusual behavior and play.[13] The prevalence of ASD in children in the general population is estimated to range from 0.6%-1%.[14,15]

Reliable data on the prevalence of ASD symptoms in children presenting with functional defecation disorders, diagnosed according to validated criteria, are sparse. In a retrospective study, 8.5% of the children had a history of diagnosed ASD with FC according to the former Rome II criteria.[16] An Australian study investigated self-reported ASD traits in young adults who had been referred for gastrointestinal symptoms in early life. No relation between gastrointestinal symptoms in early life and ASD traits in young adulthood could be demonstrated.[17] A validated assessment of the prevalence of ASD symptoms in children presenting with functional defecation disorders is needed to further clarify the possible association between the two disorders and to obtain better insight in the complex pathophysiology of both functional defecation disorders and ASD. Moreover, confirming a high prevalence of ASD symptoms in children with functional defecation disorders may give rise to an adaptation of the current standard diagnostic work-up and therapeutic strategies for functional defecation disorders in children.

ADHD	Attention deficit-hyperactivity disorder
ASD	Autism spectrum disorder
FC	Functional constipation
FI	Fecal incontinence
FNRFI	Functional non-retentive fecal incontinence
GP	General practitioner
SCQ	Social Communication Questionnaire
SCQ-L	Social Communication Questionnaire-Lifetime
SRS	Social Responsiveness Scale

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Methods

A prospective cohort study was carried out between September 2009 and October 2011 in a specialized tertiary referral center for children with functional defecation disorders at the Emma Children's Hospital, Academic Medical Centre in Amsterdam, The Netherlands. A total of 302 consecutive children, aged 4-12 years, presenting at the outpatient clinic with a diagnosis of FC or FNRFI according to the Rome III criteria were asked to participate in this study. Patients were excluded from participation if they suffered from known pathology causing constipation and/or FI, such as chronic inflammatory bowel disease, celiac disease, or when they had a history of large bowel surgery, congenital anorectal malformations, neurologic disease (complete spinal cord transection, multiple sclerosis, or spina bifida) or a genetic syndrome. Furthermore, patients with a known intellectual disability and/or an intelligence quotient of <70 were excluded. After giving informed consent, parents filled out 2 specific ASD screening questionnaires about their child. With a known prevalence of ASD in the normal population of 0.6%, a sample size of 223 children would be required to achieve a power of 80% with an alpha of 5% to detect a difference in prevalence that is at least 5 times higher than in the normal population.

This study was approved by the Medical Ethics Committee of the Academic Medical Center in Amsterdam.

Instruments

Standardized Defecation Questionnaire. During the first visit at the outpatient clinic, as part of our routine procedure a standardized defecation questionnaire was filled out by the medical doctor. This defecation questionnaire consists of questions about the medical history of the child, social environment, presence of a prior ASD diagnosis, medication use, and specific questions about the defecation pattern based on the Rome III criteria. A prior ASD diagnosis was defined as a diagnosis according to the Diagnostic and Statistical Manual of Mental Disorders criteria made by a multidisciplinary team.

Social Responsiveness Scale. The Social Responsiveness Scale (SRS) is a validated 65-item quantitative ASD screening questionnaire developed in 2005 to assess a wide range of interpersonal behavior, communication and repetitive/stereotypic behavior characteristics of ASD.[18] Items are scored on a 4-point scale, ranging from 'not true' to 'always true.' The raw total score ranges from 0-195, with higher scores indicating more ASD symptoms. In 2011, the cutoff value for raw scores in the Dutch population with normal intelligence was set at 51, with a score of ≥51 being suggestive for a diagnosis of ASD with a sensitivity of 0.90 and a specificity of 0.88 for both males and females.[19] The SRS has proven to be a reliable and valid instrument, with good psychometric properties and discriminant validity and good agreement with the Autism Diagnostic Interview–Revised.[20]

Social Communication Questionnaire-Lifetime. The Social Communication Questionnaire (SCQ) is a validated qualitative screening questionnaire for ASD in children of 4 years and older developed in 2003.[21] The SCQ consists of 40 yes or no items to be completed by the parent, resulting in a total score between 0 and 40. The agreement between SCQ scores and scores on the more extensive Autism Diagnostic Interview is high. The Social Communication Questionnaire-Lifetime (SCQ-L) version of the SCQ aims at traits of ASD in the entire developmental history of children. A total score of ≥15 on the SCQ-L is suggestive for the presence of ASD with a sensitivity of 0.90 and a specificity of 0.80.[21] The reliability and validity of the Dutch version of the SCQ-L have been approved.[22]

Outcome

The primary outcome is the prevalence of ASD symptoms in children with functional defecation disorders. The presence of ASD symptoms was defined as a score at or above the cutoff value on 1 or 2 ASD screening questionnaires (total raw SRS score of ≥51 and/or total SCQ-L score of ≥15). Scoring at or above the cutoff value on 1 of the 2 questionnaires was considered to be suggestive for the presence of an ASD. Scoring at or above the cutoff values of both questionnaires was considered to be very suggestive for the presence of an ASD. The secondary outcome is the difference in clinical characteristics between patients with and without ASD symptoms.

Statistical Analyses

For all statistical analyses, SPSS v. 16.0 (SPSS Inc, Chicago, Illinois) was used. Missing SRS values were imputed by the median value of the standardization sample; missing SCQ values by the normative (derived) score (0). A maximum of 10% missing values was considered acceptable. Total raw scores on the SRS and total raw scores on the SCQ-L were analyzed as continuous variables. For the comparison of proportions, χ^2 analyses were performed. Fisher exact tests were performed to compare proportions with an observed or expected frequency of <5. Continuous data were compared by independent t-tests when normally distributed. For the comparison of skewed continuous data, Mann–Whitney U tests were performed. The significance level was set at <0.05.

Results

Out of 302 patients, parents of 3 patients refused to participate. Three patients were excluded from participation because of a known intelligence quotient <70 and 1 because of a chromosomal abnormality (22q11 deletion) known to be associated with both constipation and ASD.[5,23]The response rate was 83%; parents of a total of 246 patients returned both ASD screening questionnaires. Because four patients had to be excluded due to too many missing values, data of 242 patients were used in the analyses. Thirty-five