

CASE REPORT

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Unexpected cause of repeated peritoneal dialysis-related complications: a case study of autism spectrum disorder with normal intelligence quotient in an adolescent

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Abstract

Background Autism spectrum disorder (ASD) is a common communication disorder, with an incidence rate of 3%. In most cases, clinicians can diagnose ASD in a single outpatient visit. However, in the case of ASD patients without intellectual disability, clinicians are sometimes unaware for a prolonged period that a patient has ASD. In such cases, delayed diagnosis can lead to serious complications.

Case presentation An 18-year-old boy had repeated severe complications of peritoneal dialysis. At the age of 9, the patient presented with proteinuria, and 5 years later, he developed end-stage kidney disease. Percutaneous renal biopsy and the clinical symptoms revealed focal segmental glomerulosclerosis with Charcot-Marie-Tooth disease due to a gene mutation in *INF2*. Peritoneal dialysis was initiated at the age of 14, but led to many related complications, including peritonitis, hypertensive retinopathy and encephalopathy, and acute heart failure. Initially, we were unaware of his developmental characteristics and autism spectrum disorder without intellectual disability, but through lengthy observations by various healthcare professionals, his unique characteristics were noticed. Because the patient often displayed a discrepancy between behavior and speech, we cautiously studied his thoughts and behaviors, and developed a special approach to ensure safe peritoneal dialysis.

Conclusions When many peritoneal dialysis-related complications occur in a patient with normal intelligence quotient, autism spectrum disorder should be considered as a possible cause.

Keywords Peritoneal dialysis, Autism spectrum disorder, Intellectual disability, Intelligence quotient, Hypervolemia, Focal segmental glomerulonephritis, Charcot-Marie-Tooth disease

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Background

Autism spectrum disorder (ASD) is a communication disorder which commonly coexists with intellectual disability. From the early developmental stage, patients develop hyperesthesia or hypoesthesia, repetitive stereotyped speech and/or behaviors, and extremely limited interest in doing any work [1]. In most cases, it is clear to general clinicians that a patient has ASD. However, in the case of ASD patients without intellectual disability (ASD-WID), which was termed Asperger disorder in the previously used Diagnostic and Statistical Manual of Mental Disorders, clinicians are sometimes unaware for a prolonged period that a patient has ASD. Here, we describe the case of an adolescent patient who had ASD with a normal intelligence quotient (IQ) and repeated serious complications of peritoneal dialysis (PD). Unawareness of ASD in this type of patient often results in severe PD-related complications due to the communication gap.

Case presentation

The present patient had a history of proteinuria revealed on a school urinalysis screening at the age of 9 years. By the age of 13, his urinary protein creatinine ratio gradually increased to 9.04 and the estimated glomerular filtration rate dropped to 46.4 mL/min/1.73 m². Ultrasound-guided percutaneous renal biopsy, genetic testing, and steppage gait revealed that he had hereditary focal segmental glomerulosclerosis with Charcot-Marie-Tooth disease, caused by a gene mutation in the *INF2* (c.218G>T). End-stage kidney disease developed rapidly and PD was initiated at the age of 14. During the first PD catheter insertion and subsequent educational hospitalization, he experienced PD-related peritonitis due to unhygienic practices. This complication was caused by continuation of the routine PD procedure while playing a TV game, even though he noticed dialysate leaking from the PD tube. Around the same time, he began to feel difficulty moving his right leg. His parents both worked full-time and were often not at home by the time automated peritoneal dialysis was initiated. Therefore, although his parents had successfully learned how to correctly perform PD procedures, the patient needed to do the same so that he could perform them completely on his own at home.

Within the next 4 years, the patient was hospitalized five times due to hypervolemia, hypertensive retinopathy, seizure clusters from hypertensive encephalopathy, dehydration, and acute heart failure. Although he experienced many complications related to hypervolemia, he still desired to gain weight through drinking water as he perceived the loss of more weight than his classmates as a sign of frailty. During a hypertensive encephalopathy

episode at the age of 15, he presented with drop foot in both legs. Along with the results of a neuroelectric examination, he was diagnosed with Charcot-Marie-Tooth disease. After the hypertensive encephalopathy episode, the attending doctor warned that additional self-inflicted PD-related complications would inhibit him from continuing PD, and he would require hemodialysis. Not wanting to receive hemodialysis, he started weight restriction for half a year. Consequently, he was hospitalized for dehydration, triggered by gastroenteritis. This dehydration episode caused discomfort, leg cramps, and insomnia, and he began to gain weight again. At this time, he realized that appropriate weight control could be achieved through equating the amount of drinking water to the amount of fluid removed by PD. This assumption did not include the fluid intake of each meal, and subsequently led to hypervolemia and acute heart failure.

During hospitalization for the latter, we noticed that the patient had ASD. After 3 months, he was hospitalized again with PD-related peritonitis due to unhygienic practices. Although difficult, he endured peritonitis pain for a week before hospitalization. We suggested that he take a reasoning test, the Wechsler Adult Intelligence Scale, third edition. His Full-Scale IQ was normal at 109, individually he had a Verbal IQ of 120, Performance IQ of 92, Working Memory of 137, and a Processing Speed of 78, showing large differences between these IQs. We began a multidisciplinary treatment intervention conducted by multi-specialty medical professionals, including pediatric nephrologists, urologists, dialysis physicians, psychiatrists, senior and junior residents, nurses, pharmacists, psychologists, nutritionists, physical therapists, and medical representatives. For example, all staff worked together to create a checklist card of PD procedures specifically for him. During hospitalization, the patient performed the PD procedures himself, and the nurses reviewed them and immediately pointed out any mistakes so that he could continue the correct PD procedures at home. Our other special approaches adopted to manage his traits are indicated in Table 1. Following the lengthy period required to explain his personality traits and treatments, the patient recognized his disease better than before. His therapeutic behavior towards self-controlled PD has changed and continues to improve.

Discussion and conclusions

Why did the patient receive PD, not kidney transplantation?

Based on the presence of multiple complications associated with PD, kidney transplantation might be considered as the first treatment choice. However, in Japan, organ donation is infrequent and patients often wait for several years before receiving a kidney transplant [2]. Therefore,

Table 1 The patient's superficial behaviors, his interpretation, and our responses

The patient's superficial behaviors	Interpretation of behaviors as symptoms of ASD	Our approach
Hurling abuse at healthcare professionals	Communication disorder	Avoid irritation
Cannot read someone's face	Unsmooth non-verbal communication	Using more verbal communication
Cannot maintain PD records	Limited scope of interests	Using a smartphone instead of a notebook
Obsession with food	Limited scope of interests	Allow preferred meal once a week
Cannot remember someone's name	Limited scope of interests	Introducing ourselves every time
Endured peritonitis pain for a week	Hypoesthesia	Checking the patient's general condition over the phone daily
Unhygienic PD procedure	Obsession with own procedure	Following the checklist of standard PD procedure
Incorrect assumptions about water restriction	Obsession with own idea	Explaining correct theory repeatedly

ASD autism spectrum disorder, PD peritoneal dialysis

undergoing PD while awaiting kidney transplantation is a common course of treatment for children in Japan.

Taking the present patient's repeated major complications into account, hemodialysis might be considered a better treatment option. However, PD procedures can be performed, the PD schedule can be changed in accordance with the body weight, the mechanism of action of the prescription can be explained, and the importance of dietary restrictions can be understood by the patient. Therefore, PD was decided as the optimal option.

Problematic behaviors/characteristics of ASD observed in the patient

Some clinicians had reported that they could not manage the patient's treatment-related behaviors because he often lies argumentatively [3, 4]. Furthermore, it was noted that often neglected aseptic principles and safety while performing PD [5]. This was because the patient dislikes following the official manuals and prefers to perform PD procedures hastily. The patient often drinks more water than indicated because he thinks it can be easily removed with higher-glucose concentration PD dialysate. He tends to consume too much sodium because he is obsessed with meat and junk food. As people with ASD-WID have poor recollection of names and figures of people and objects [6], he also had trouble remembering the prescription, and therefore often did not follow it.

What healthcare professionals can do for ASD patients with normal IQ

We noted that the patient often lies without intending to. Therefore, it is difficult for healthcare professionals to determine discrepancies between his words and actions [7]. For example, when clinicians ask the patient to show his PD recorded notebook, he always says that he does not have it with him, although the real reason was that he has not updated it. The cognition of patients

with ASD-WID tends to be distorted [8]; if the PD record notebook is out of the patient's field of vision, he may forget to update it. As he always had his smartphone with him, we suggested maintaining a record of PD details using an application on his smartphone, and he started continuous recording thereafter, and he was subsequently able to present the results to the healthcare professionals upon request. Thus, patients with ASD-WID require unique administration of PD treatment to avoid life-threatening PD-related complications [9, 10], though this does not mean that these patients are unmotivated to undergo treatment. When certain severe PD-related complications occur in a normal IQ patient, it is important for all clinicians and various other healthcare professionals to reconfirm whether the patient has underlying ASD [11].

The communication gap between healthcare professionals and patients with ASD commonly results in severe medical complications. If patients with PD-related complications frequently act inappropriately, beyond the clinicians expectations, we should carefully observe them with the help of various medical specialists. The cognition of patients with ASD-WID is distorted, and special aids may significantly improve patient condition, for example in establishing the safety of PD procedures.

Mini-review

ASD and ASD-WID

ASD is a well-known communication disorder, in which patients have characteristic obsessions and behaviors. In the latest Diagnostic and Statistical Manual of Mental Disorders fifth edition of the American Psychiatric Association [1], the definition of ASD was broadened to emphasize that it is a spectrum. This means that different patients have various degrees of ASD, with some ASD patients not manifesting communication problems [12]. Diagnosis of a typical ASD patient is generally easy

for general clinicians. However, diagnosis of an ASD patient whose communication skills appear to be normal is often more complicated. Although the percentage of ASD-WID varies in each report, many ASD patients have intellectual disability [13]. Diagnosis of ASD patients with intellectual disabilities is often easier than that of ASD-WID. This is partly because some ASD symptoms overlap with symptoms of intellectual disability, as well as because some communication problems in ASD-WID patients can be overcome, for example, by pattern and repetitive learning. Therefore, accurate communication to convey therapeutic behaviors with ASD patients with a normal range of communication skills and normal IQ can be very difficult in routine medical interviews.

Abbreviations

ASD	Autism spectrum disorder
ASD-WID	Autism spectrum disorder without intellectual disability
IQ	Intelligence quotient
PD	Peritoneal dialysis

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Consent for publication

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Competing interests

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