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BRIEF REPORT

Brief Report: Implementation of a Specific Carbohydrate Diet for a Child with Autism Spectrum Disorder and Fragile X Syndrome

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Abstract

This brief report examines the implementation of dietary intervention utilizing the specific carbohydrate diet (SCD) for the management of gastrointestinal issues in a 4 year old boy diagnosed with Autism Spectrum Disorder (ASD) and Fragile X Syndrome (FXS). Data relating to anthropometrics, dietary intake, blood markers, gastrointestinal (GI) symptoms, sleep issues, and behavioral concerns were gathered at baseline and after 4 months of dietary intervention. The dietary intervention was well tolerated. Improvements in nutrient status, GI symptoms, and behavioral domains were reported. The use of the SCD protocol in children with ASD/FXS and GI symptoms warrants further investigation.

Keywords Autism spectrum disorder · Fragile X syndrome · Nutrition · Specific carbohydrate diet

Introduction

Autism spectrum disorder (ASD) is a complex pervasive developmental disorder characterized by impairments in social interaction, with deficits in verbal and non-verbal communication and/or restricted repetitive and stereotyped patterns of behavior and interests (American Psychiatric Association 2013). A high proportion (46–84%) of children with ASD are reported to have gastrointestinal (GI) dysfunction and associated symptoms, including, but not limited to, gastroesophageal reflux disease (GERD), abdominal pain, constipation, and diarrhea (Buie et al. 2010a, b; Black et al. 2002; Valicenti-McDermott et al. 2006; Holingue et al. 2017). Associated functional GI abnormalities in children with ASD also include low activities of disaccharidase enzymes (Melmed et al. 2000; Williams et al. 2011), defective sulfation of ingested phenolic amines, such as acetaminophen (Alberti et al. 1999), bacterial overgrowth with a generalized reduction in biodiversity (Kang et al. 2013; Wang et al. 2017), and increased intestinal permeability (D'Eufemia et al. 1996; Williams et al. 2011). Severe GI symptoms in children compromise dietary intake and can cause nutritional depletion (Hartman et al. 2009). Furthermore, because children with ASD are often nonverbal, vigilance is needed on the part of the treating physician to identify underlying chronic medical conditions, such as GERD, that can contribute to failure to thrive, irritability, and/or behavioral concerns.

Fragile X syndrome (FXS) is the most common known genetic cause of inherited intellectual disability and the most common known single-gene cause of ASD (Hagerman et al. 2011). Children with FXS experience a wide range of medical problems, including cardiac concerns, otitis media, seizures, sleep disorders, and gastrointestinal symptoms (Kidd et al. 2014). The frequency of GI disorders in children with FXS has not been well studied but is thought to be around 11%. Hypotonia and connective tissue abnormalities are thought to contribute to GI problems, including GERD, irritable bowel syndrome, constipation, and diarrhea resulting in inadequate nutrition and increased risk for failure to thrive syndrome (Goldson and Hagerman 1993).

The specific carbohydrate diet (SCD) was developed in the 1930s as a dietary protocol intended for patients with celiac disease (Haas and Haas 1955) but has since been employed to treat Crohn's disease, ulcerative colitis, diverticulitis and chronic diarrhea (Galland 1999; Gottschall 2004; Suskind et al. 2014; Obih et al. 2016). The SCD

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protocol calls for strict avoidance and elimination of all grains, lactose, and sucrose derived carbohydrates from a person's diet with the direct aim of restoring and maintaining a healthy intestinal microbial profile [see (Gottschall 1994) for detailed dietary information]. While bacteria in the intestinal tract naturally occur, and are, in fact, necessary for healthy and adequate digestive function, an overabundance of any single bacterial species can have profound effects including: (i) the production of an excess of short chain organic acids (thus lowering the pH of the colon); (ii) an increase in the production of metabolic byproducts of fermentation as well as bacterial toxins; and (iii) a possible mutation of some harmless bacteria into pathological forms (Gottschall 1994; Lane et al. 2017; Albenberg and Wu 2014; den Besten et al. 2013). Such buildups have been associated with epithelial inflammation of the intestinal mucosa, malabsorption of vitamin B12 in the ileum, and lipid malabsorption in the ileum and jejunum, which could perpetuate deficiencies in fat soluble vitamins (Brandt et al. 1977; Bures et al. 2010; Kau et al. 2011; DiBaise 2008). Bacterial overgrowth may also damage intestinal villi, limiting the surface area within the intestine that is capable of absorbing nutrients properly resulting in gas, bloating, abdominal cramping, diarrhea and steatorrhea (DiBaise 2008).

Implementation of the SCD protocol in children with ASD with gastrointestinal concerns has not been evaluated and yet it is an intervention that is widely used with and without clinical guidance by many families. This brief report evaluated whether the application of a nutrient dense SCD in a child with ASD FXS, and significant GI symptoms was well-tolerated and met nutritional demands, such that GI symptoms were ameliorated resulting in increased growth velocities and improvements in aberrant behaviors.

Methods

Screening Procedures

This brief report describes data obtained from a 4 year old boy recruited as part of a larger dietary intervention study. The participant's mother underwent a pre-screening via phone to determine the child's initial eligibility for the study. Inclusion criteria were as follows: 2–6 year old girls and boys with ASD or with suspected ASD, as well as evidence of GI symptoms including diarrhea, constipation, abdominal pain, failure-to-thrive, malodorous stool, blood or mucus in the stool, and/or GERD. In addition, the child's parent/caregiver had to be willing to participate by completing the necessary measures and strictly following all dietary interventions, and be accessible to a clinician via phone or in-office on a weekly basis. No major changes could be made to any adjunct behavioral therapies that the child was receiving during the 16 week SCD intervention. Exclusion criteria were as follows: previous SCD intervention; diagnosed metabolic disorders such as phenylketonuria, leukodystrophy, lysosomal disorder, and Wilson's disease; diagnosed genetic syndromes such as Down's Syndrome, Angelman Syndrome, Prader-Willi, Rett's Syndrome, and FXS; other concurrent physical, mental or neurological disorders that preclude participation in assessment procedures; requirement of receiving any nutrition via enteral (tube) or parenteral (intravenous) routes; use of any psychiatric medications (or psychiatric medications within the last 3 months prior to enrollment); and not a participant in a concurrent, formal therapeutic trial. The mother expressed interest in the study, the informed consent form was reviewed with her, and consent obtained.

During the screening process, the child's mother did not indicate that their child had any genetic conditions that would exclude them from participating in this study, even though this question was specifically asked. The child was enrolled in the study and progressed through the study procedures as planned. At the participant's 6 month study visit, we received additional paperwork from the parents that included a genetic evaluation conducted when the participant was 3 years of age indicating that the participant had a double mutation on the COMT gene, mutations on Chromosome 16 (16p11.2) and the X chromosome, and been diagnosed with FXS. The participant was therefore removed from the study at that time. This single case of a child with both an ASD and FXS diagnosis is therefore presented herein.

Study Procedures

This was a longitudinal study conducted over approximately 6 months. Behavioral, dietary, biochemical and GI assessments were conducted at baseline prior to SCD implementation, and then again after 16 weeks of dietary intervention.

Diagnostics

A previous diagnostic assessment conducted by a licensed psychologist within the prior year including administration of the ADOS-2 and ADI-R indicated that the subject met the classification of ASD. Additional testing was therefore not conducted at enrollment.

Anthropometrics

A nutrition-based physical exam was conducted by a licensed, registered dietician at baseline and after 16 weeks of SCD intervention. BMI was calculated as kg/m^2. Height and weight were compared with the standards for linear growth derived from the Centers for Disease Control-National Center for Health Statistics Growth Charts

(Institute of Medicine 2005). Other anthropometrics collected included triceps skinfold (TSF) and mid-arm circumference (MAC), from which, a mid-arm muscle circumference (MAMC) was calculated as an indicator of protein and fat reserves.

Nutritional Assessment

At the time of enrollment in the study, the participant had been on a gluten- and dairy-free (GFCF) diet for almost 24 months. The family initiated this GFCF intervention at this time without professional recommendation or guidance, using information provided by other parents to support them in that process. Approximately 6 months later, they sought clinical support from a registered dietician for assistance with the GFCF diet. A 3-day food diary was conducted by the mother at baseline and after 16 weeks of dietary intervention to record all food and beverage intake for 2 week days and 1 weekend day that they considered typical for their child (Mari-Bauset et al. 2016). Food records were analyzed using Food Processor SQL (version 10.3.0, 2008, ESHA, Salem, OR). Data for the 3 days were combined using the Multiple Source Method (Haubrock et al. 2011). The participant was on a multi-vitamin and essential fatty acid supplement. Supplement data was not included in the nutrient analysis for an accurate assessment of dietary nutriture.

Biochemical Measures

A fasting blood sample was obtained at baseline and 16 weeks and processed for a comprehensive metabolic panel (CMP), complete blood count (CBC), C-Reactive Protein (CRP), and liver and pancreatic function tests by Clinical Pathology Laboratories, Inc. (CPL).

Gastrointestinal Assessment

An on-line gastrointestinal symptoms questionnaire (GSQ) was developed in-house and included questions about stool characteristics (size, form, frequency, odor, color etc.) and the child's behavior (hyperactivity, irritability, straining to pass a bowel movement, reflux, gagging, spitting etc). Yes/no scores were scored 1 or 0. Descriptive data scores were weighted 0, 1, or 2 based on the severity of response. All scores were then summed to obtain a composite score. The GSQ was completed at baseline, and daily throughout the study until 16 weeks, then weekly thereafter.

Behavioral Assessment

The pervasive developmental disorder behavior inventory [PDDBI; (Cohen et al. 2003)], a validated rating scale for assessing response to intervention in children with a pervasive developmental disorder, such as ASD, was conducted at baseline and at 16 weeks. Problem behaviors measured included stereotyped behaviors, fears, aggression, social interaction deficits, and aberrant language.

Sleep Assessment

The child sleep history questionnaire (CSHQ), a 45-item parent questionnaire that examines sleep behavior in young children (Owens et al. 2000), was implemented at baseline and 16 weeks post intervention to assess changes in sleep patterns over time.

Implementation of SCD

All food that the participant consumed over the 16 weeks of SCD intervention was prepared by a private chef using local and organic food sources and shipped directly to the parents. Three meals and two snacks per day were provided for 16 weeks. These shipments included a menu plan of foods designed specifically within the requirements of an SCD intervention for the child's food preferences. Meal options for this participant included a variety of prepared protein such as chicken nuggets, grilled chicken breasts, ground bison taco meat, and wild Atlantic salmon filets; fruits such as peaches, pears, berries, and prepared applesauce; vegetables such as carrots, butternut squash, spinach, and green beans; banana bread muffins; and SCD compliant ice cream. Organic pear juice was included in these shipments so that daily consumption of up to 16 oz could be sustained. It was recommended that water replace all other beverages with the exception of the pear juice. This food was provided at no cost to the family. The child did not consume any additional food or drink outside of the prescribed foods provided as part of this study.

A dietician monitored the implementation and adherence to the intervention and prepared menus tailored for the participant to ensure that the participant's nutritional status remained uncompromised. The SCD protocol was tailored, taking into consideration the participant's food preferences and nutritional needs. Foods not supported in the SCD protocol were gradually excluded over the first week of the initiating study intervention. As the participant's food preferences expanded to meet the criteria for the SCD, the dietician's recommendations included an increase in nutrient dense foods meeting the criteria outlined by the SCD. Weekly counseling was provided and nutritional support was offered to ensure that the child had a nutritionally adequate intake, with the appropriate number of calories and proper nutrient density.

Results

Anthropometrics

Changes in the participant's anthropometric measures from baseline to 16 weeks of SCD intervention were as follows: weight increased from 43 lb (90th percentile) to 46 lb 8 oz (88th percentile); height increased 42.5in (90th percentile) to 43.5 in (75th percentile); and BMI increased from 16.7 (81st percentile) to 17.3 (90th percentile). At baseline, MAC was 180 mm, TSF was 11 mm, and MAMC was 145.46 mm placing the participant between the 50th and 75th percentiles for his age. At the end of the 16 weeks of SCD intervention, MAC was 187.5 mm, TSF was 13.5 mm, and MAMC was 145.11 mm, which placed him between the 50th and 75th percentiles for his age.

Nutritional/Biochemical Assessments

At baseline, an analysis of a 3-day food diary revealed appropriate intake of macronutrients and low intake of vitamins D (34% of RDA) and E (66% of RDA), calcium (28% of RDA), iodine (13% of RDA), choline (57% of RDA), molybdenum (69% of RDA) and potassium (70% of RDA). The participant's daily intake met or exceeded the RDA for all other micronutrients. A similar analysis, after 16 weeks of SCD intervention, indicated increased protein intake (859% of RDA), decreased carbohydrate intake (62% of RDA), and low intakes for vitamin D (70% of RDA), calcium (45% of RDA) and iodine (68% of RDA), although intakes for these three micronutrients had all increased considerably over baseline levels (Table 1). The participant's daily intake met or exceeded the RDA for all other macro- and micronutrients. Lab work completed at baseline and at 16 weeks was unremarkable.

Table 1 Comparison of daily nutrient intake before and after 16 weeks implementing the SCD protocol

	Baseline			16 weeks			% Change[a]
	Value	RDA	% of RDA	Value	RDA	% of RDA	
Macronutrients							
Energy (kcal)	1837.29	1746.11	105	2544.10	1819.98	140	35
Protein (g)	70.82	18.96	374	244.33	19.82	1233	859
Carbohydrates (g)	231.41	240.09	96	154.95	250.25	62	−34
Total fat (g)	69.53	54.32	128	107.43	56.62	190	62
Fiber (g)	23.18	24.45	95	30.94	25.48	121	26
Micronutrients							
Vitamin A RAE	1187.11	400.00	297	3256.67	400.00	814	517
Vitamin C (mg)	153.43	25.00	614	98.35	25.00	393	−221
Vitamin D (mcg)	5.07	15.00	34	10.47	15.00	70	36
Vitamin E (mg)	4.65	7.00	66	12.92	7.00	185	119
Vitamin B1 (mg)	0.85	0.60	142	1.38	0.60	230	88
Vitamin B2 (mg)	0.79	0.60	122	2.45	0.60	408	286
Vitamin B3 (mg)	14.52	8.00	182	45.12	8.00	564	382
Vitamin B6 (mg)	1.46	0.60	243	1.46	0.60	728	485
Folate—DFE (mcg)	278.57	200.00	139	500.10	200.00	250	111
Vitamin B12 (mcg)	2.46	1.20	205	13.98	1.20	1165	960
Calcium (mg)	279.64	1000.00	28	449.46	1000.00	45	17
Iron (mg)	21.02	10.00	210	21.02	10.00	233	23
Magnesium (mg)	153.03	130.00	118	389.53	130.00	300	182
Zinc (mg)	5.1	5.00	102	26.91	5.00	538	436
Trace minerals							
Iodine (mcg)	11.86	90.00	13	11.86	90.00	68	55
Choline (mg)	141.67	250.00	57	804.36	250.00	322	265
Selenium (mcg)	33.97	30.00	113	214.10	30.00	714	601
Manganese (mg)	1.8	1.50	120	2.49	1.50	166	46
Molybdenum (mcg)	15.28	22.00	69	35.31	22.00	161	92
Potassium (mg)	2657.31	3800.00	70	488.09	3800.00	129	59

RDI recommended daily allowance

[a]Represents the change in daily nutrient intake from baseline to 16 weeks

Gastrointestinal Symptoms

The participant experienced marked change in GSQ composite score. Beginning at a baseline of 11 (primarily due to stool size, consistency, texture, the amount of strain that was exhibited by the participant to pass stool, and the need for an enema). After 16 weeks of SCD implementation, the GSQ composite score had improved to a score of 3 (primarily due to stool consistency and level of irritability). The biggest difference in gastrointestinal symptoms that the parents reported after SCD implementation was that the child was able to have a bowel movement without considerable straining and the requirement of a suppository or enema.

Table 2 Comparison of CSHQ sleep domains before and after 16 weeks implementing the SCD protocol

Domain	Baseline	16 weeks	Change in score[a]
Bedtime resistance	6	8	+2
Sleep onset delay	1	1	0
Sleep duration	3	4	+1
Sleep anxiety	4	6	+2
Night wakings	3	3	0
Parasomnias	9	9	0
Sleep disordered breathing	3	3	0
Daytime sleepiness	14	9	−5
Total sleep disturbance score	43	43	0

[a]Improvements in CSHQ domains are indicated by a negative change in score

Sleep Assessment

The participant's baseline score on the CSHQ was 43 (Table 2), higher than the cut-off point suggestive of sleep problems established for the CSHQ (Owens et al. 2000). While this score was unchanged following SCD intervention, some domains increased slightly, whereas some decreased (Table 2). The biggest change reported was in day-time sleepiness decreasing from a score of 14 to 9.

Behavioral Assessments

Changes were noted in the overall PDDBI autism composite T score, decreasing 15% (from 60 to 51) from baseline to the end of SCD intervention. This was primarily driven by a reduction in sensory, repetitive, and ritualistic behaviors, semantic/pragmatic problems, and specific fears and an increase in receptive and expressive language, and learning and memory, as shown in the reported composite scores (Table 3). The patient did not start on any new medications during the 16-week intervention, and no changes were made to his medication regimen or behavioral therapies during this time frame.

Table 3 Comparison of PDDBI domain and composite scores before and after 16 weeks implementing the SCD protocol

Domain/composite	Baseline (T0)			16 weeks (T5)			% change T score[a]
	Raw score	T score	90% CI	Raw score	T score	90% CI	
Approach/withdrawal problems							
Sensory/perceptual approach behaviors	21	51	45–57	15	48	42–54	6
Ritualisms/resistance to change	17	59	52–66	14	51	44–58	14
Social pragmatic problems	21	64	56–72	23	60	53–67	6
Semantic/pragmatic problems	30	76	70–82	19	58	51–65	24
Arousal regulation problems	18	51	43–59	20	54	47–61	−6
Specific fears	37	68	62–74	29	59	53–65	13
Aggressiveness	42	80	75–85	38	75	68–82	6
Repetitive, ritualistic, and pragmatic problems composite	89	67	61–73	71	56	51–61	16
Approach/withdrawal problems composite	184	71	66–76	158	62	58–66	13
Receptive/expressive social communication abilities							
Social approach behaviors	75	61	57–65	74	53	49–57	13
Expressive language	53	59	56–62	89	63	60–66	−7
Learning, memory, and receptive language	20	53	49–57	34	58	54–62	−9
Expressive social communication abilities composite	128	61	58–64	163	60	57–63	2
Receptive/expressive social communication abilities composite	148	59	57–61	197	60	58–62	−2
Autism composite	130	60	56–64	101	51	47–55	15

[a]Improvement in the approach/withdrawal problems domain is indicated by a positive percent change in T score. Improvement in the receptive/expressive social communication abilities domain is indicated by a negative percent change to T scores

Discussion

This is the first study examining the implementation of an SCD protocol in a child with ASD and FXS. While the effects of the SCD protocol implementation in individuals with neurodevelopmental disorders is an area that warrants further research, the results in the case of the child presented, suggest that the SCD protocol was well tolerated and met nutritional demands. Improvements in gastrointestinal health, and behavior were reported following 16 weeks of strict adherence to this intervention.

In children with ASD, there have been contradictory reports on growth measurements across several studies [reviewed by (Mari-Bauset et al. 2014a)], although it appears that in one Spanish study, children with ASD had a lower BMI compared with healthy controls (Mari-Bauset et al. 2012). In children with FXS, weight and height in males closely approximate to normal percentiles until about 12–15 years of age, when FXS weight percentiles are above the norms and height percentiles fall below the norms (Butler et al. 1992). In this case report, the participant's height and weight increased appropriately from baseline to the completion of dietary intervention resulting in a slight increase in BMI. It has been proposed that implementation of an SCD protocol in people with celiac disease restores the balance of bacteria within the GI tract thereby resolving the associated dysbiosis allowing for greater access to and absorption of nutrients (Haas and Haas 1955), which would likely result in increased weight gain. Weight gain after implementing an SCD has been documented in several studies on IBD (Braly et al. 2017; Haskey and Gibson 2017). Muscle measurements remained similar in the 50th to 75th percentiles suggesting that growth measures were not negatively impacted by the 16 week SCD intervention.

Many children with ASD have feeding behaviors that result in picky eating and food selectivity (Mari-Bauset et al. 2014b), often resulting in poor, unbalanced diets high in processed foods and saturated fats (Mari-Bauset et al. 2015). Children with ASD tend to consume fewer calories, more carbohydrates, and less protein and fat than typically developing controls (Neumeyer et al. 2018). While there is a paucity of information regarding nutrient status in FXS, increased folic acid, and essential fatty acids may be helpful (Rueda et al. 2011; Pietropaolo et al. 2014). Many children with ASD and/or FXS are on restricted diets (Rubenstein et al. 2018), which may give rise to potential nutritional deficiencies (Arnold et al. 2003). However, with suitable clinical support, dietary intake need not be adversely affected (Barnhill et al. 2017; Cornish 2002). The participant in this case study had been on a gluten-free and dairy-free diet for almost 24 months and under the care of a registered dietitian for almost 18 months prior to enrolling in this study, and his baseline nutrient intake met RDA for the majority of macro- and micronutrients. Additionally, the participant's food preferences changed little over the course of the intervention. He was not considered a selective or picky eater at the time of study initiation. Over the preceeding 2 years he had been exposed to a variety of healthy foods, and worked with a registered dietician and a team of ABA therapists who all supported his nutritional care. In this time frame, gluten and casein were eliminated from his diet and he moved from a highly processed, typical American diet 2 years prior to study initiation to the one outlined in Table 4. The impact of his prior GFCF status cannot be quantified for the purposes of this study, but one can surmise that his acceptance of new foods made the transition to an SCD protocol an easier process for the family.

When implementing an SCD protocol, however, it is anticipated that carbohydrate intake will decrease, and be compensated for by increased protein and fat intake (Braly et al. 2017). Protein intake in the participant described in this case study increased considerably after implementing the SCD to levels greater than the current RDA. However, high-protein diets lead to increased intestinal absorption of calcium, increased levels of circulating insulin-like growth factor-1, and decreased serum parathyroid hormone level (Vatanparast et al. 2007). In a recent study, the intake of higher animal protein, calcium and phosphorus were positively associated with bone density measures in children with ASD leading the authors to conclude that children with ASD should focus on higher protein intakes than the RDA (Neumeyer et al. 2018).

Children with ASD frequently report GI concerns, including GERD, diarrhea, constipation and pain (Holingue et al. 2017). In FXS, GERD, constipation, and diarrhea are most commonly reported and are thought to be associated with hypotonia and connective tissue abnormalities (Kidd et al. 2014). In this case report, the child had a number of abnormal stool characteristics including grainy, foul-smelling stool that contained undigested food. It was also reported that the child demonstrated severe straining to try to pass a bowel movement, and typically, this was not successful without the aid of a suppository or enema. Despite being on a GFCF diet prior to study enrollment, the GI issues for the participant were a significant concern. Following 16 weeks of SCD intervention, the child's bowel movements were no longer foul-smelling nor did they contain undigested food. In addition, the level of straining to pass a bowel movement had decreased and medical support through suppositories or enemas was no longer necessary demonstrating an overall improvement in gastrointestinal symptoms following implementation of the SCD protocol. Improvements in clinical symptoms after SCD intervention have also been reported

Table 4 Typical daily intake for participant

Meal	T0—baseline	T5—intervention week 16
Breakfast	2 4-inch pancakes, prepared from gluten free pancake mix (using unsweetened almond milk and organic non-dairy spread) 2 pieces organic bacon 12 blueberries Water	1 large egg, fried 3 oz chicken sausage links 15 blueberries 1 SCD homemade banana bread muffin 8 ounces pear juice
Morning snack		1 medium apple Water
Lunch	2 quesadillas prepared with rice flour, non-dairy cheese substitute, and ground beef 6 raspberries 12 cherry tomatoes Water	6 ounce beef hamburger patty 15 baby carrots 1/2 cup fresh spinach water
Afternoon snack	1 banana 12 gluten free pretzels 6 slices cucumber Water	1 banana Water
Dinner	2 quesadillas, prepared with: rice flour, 3/4 cup shredded chicken, 1 sliced red bell pepper, and 1 mushroom Water	1 cup fresh spinach 7 baby carrots 6 ounces butternut squash cubes, baked 14 ounces mixed wild caught atlantic salmon, baked and atlantic cod 8 ounces pear juice
Evening snack	12 blueberries 6 slices cucumber 1 beef/pork blend sausage link, 3 ounces Water	1/2 cup unsweetened applesauce 5 SCD chicken nuggets 5 baby carrots Water

in pediatric patients with Crohn's Disease and IBD (Suskind et al. 2014, 2016).

Children with ASD often report problems with sleep including bedtime resistance, difficulty falling asleep, and nighttime wakening (Johnson et al. 2012). In FXS, the most commonly reported sleep concerns include difficulty falling asleep and nighttime wakings (Richdale 2003). The CSHQ can assist in identifying co-morbid sleep disturbances that may complicate the presentation of underlying medical or mental health concerns in children (Owens et al. 2000). In this study, the participant's sleep disturbance score at baseline met the cut-off for sleep concerns but the score this remained relatively constant following SCD intervention for 16 weeks. The biggest change reported was for daytime sleepiness, which decreased 5 points following SCD intervention. This may be correlated with fewer GI symptoms, which can cause disrupted sleep in children with ASD (McCue et al. 2017).

Behavioral and cognitive symptoms reported in FXS have some overlap with ASD, including poor eye contact, perseveration in speech and behavior, and tactile defensiveness, suggesting an increased sensitivity to a variety of stimuli (Hagerman 1991). There is also an aberrant approach-withdrawal behavioral pattern in social interactions in FXS (Cohen et al. 1989). In this subject, we identified a reduction in maladaptive behaviors such as sensory, repetitive, and ritualistic behaviors, semantic/pragmatic problems, and specific fears, and an increase in adaptive behaviors such as receptive and expressive language, and in learning and memory following 16 weeks of SCD intervention. Overall, these changes led to a 15% decrease in the PDDBI Autism Composite score corresponding to an overall improvement in ASD-related symptomatology (Cohen and Sudhalter 2005). The participant was also receiving 16–20 h of applied behavioral analysis (ABA) therapy over the course of this time frame, which did not change from study initiation.

To summarize, a 16 week intervention following the SCD protocol was well tolerated in this 4 year old child diagnosed with ASD and FXS, leading to improvements in growth status, gastrointestinal symptoms, and behaviors. Further research is needed to further evaluate implementation of the SCD protocol in young children with ASD and/or FXS and GI concerns.

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Compliance with Ethical Standards

Conflict of interest All of the authors declare that they have no conflicts of interest.

Ethical Approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from the participant's representative according to the above-mentioned principles.

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