RESEARCH ARTICLE

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Contributions of a Specialty Clinic for Children and Adolescents with Down Syndrome

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We investigated what added value, if any, a Down syndrome specialty clinic brings to the healthcare needs of children and adolescents with Down syndrome. For this quality improvement study, we performed a retrospective chart review of 105 new patients with Down syndrome, ages 3 and older, seen during the inaugural year of our specialty clinic. We asked how many of our patients were already up-to-date on the healthcare screenings recommended for people with Down syndrome. We further analyzed what tests we ordered, which referrals we suggested, and, ultimately, what new diagnoses of co-occurring medical conditions were made. Only 9.8% of our patients were current on all of the recommended Down syndrome healthcare screenings. Parents came to clinic with a variety of concerns, and after laboratory tests, radiologic studies, and subspecialty referrals, we made many new diagnoses of gastrointestinal conditions (e.g., constipation and celiac disease), seasonal allergies, dermatologic conditions (e.g., xerosis), behavioral diagnoses (e.g., autism spectrum disorder and disruptive behavior not otherwise specified), and clarifications of neurologic conditions. A Down syndrome specialty clinic can identify and address many healthcare needs of children and adolescents with Down syndrome beyond that which is provided in primary care settings.

Key words: Down syndrome; ambulatory care; clinical practice; trisomy 21

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INTRODUCTION

The approximately 250,000 people with Down syndrome (DS) living in the United States [Presson et al., 2012] are predisposed to numerous medical conditions, many preventable and most treatable, yet only 58 DS specialty clinics exist in 32 states [National Down Syndrome Society, 2012a]. Where clinics are absent, primary care physicians have typically been charged with coordinating the comprehensive medical care; and since the 1970s, published recommendations have offered some direction. Over the years, however, the guidelines formalized by the American Academy of Pediatrics (AAP) and the Down Syndrome Medical Interest Group (DSMIG), have grown longer and more complex [Van Cleve and Cohen, 2006; Van Cleve et al., 2006; Bull and Committee on Genetics, 2011; Cohen, W.I. for the Down Syndrome Medical Interest Group, 1999]. An open question has emerged: is it reasonable and practical for the primary care physician to continue to coordinate such care in an era where medicine is often rewarded for time efficiency?

One of the long-standing recommendations in all DS healthcare guidelines in the United States is that people with DS should have their thyroid function tests checked annually, as 4–18% of people with DS can develop hypothyroidism, hyperthyroidism or hyperthyrotropinemia throughout their lives [Bull and Committee

Abbreviations: DS, Down syndrome; AAI, atlantoaxial instability; OAI, occiptoaxial instability; AAP, American Academy of Pediatrics; DSMIG, Down Syndrome Medical Interest Group; CBC, complete blood count; IEP, individualized education plan; ABA, applied behavioral analysis; BMI, body mass index.

Conflict of Interest: Brian Skotko serves in a non-paid capacity on the Board of Directors for the not-for-profits Massachusetts Down Syndrome Congress and Band of Angels Foundation. He also serves in a non-paid capacity on the Professional Advisory Council of the not-for-profit National Down Syndrome Congress. Dr. Skotko occasionally gets remunerated from Down syndrome non-profit organizations for speaking engagements about Down syndrome. He has a sister with Down syndrome.

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on Genetics, 2011]. Between 1994 and 2004, <15% of children with DS living in Oklahoma and Nebraska were found to be up-to-date on such annual screens [Fergeson et al., 2009]. In a separate study at the University of Michigan, <50% of adults with DS were evaluated for obstructive sleep apnea, atlantoaxial instability, hearing loss, and vision loss, all screening recommendations with broad consensus for improving health outcomes [Jensen et al., 2012].

DS specialty clinics have been created to ensure that people with DS remain current on healthcare screenings, receive attention for developmental and behavioral concerns, and access coordinated, comprehensive care. Some clinics function in a primary care capacity exclusively for patients with Down syndrome; others serve as tertiary referrals centers, situated most commonly in divisions of genetics, developmental–behavioral pediatrics, neurodevelopmental disabilities or neurology.

Since 1967, the Down Syndrome Program at Boston Children's Hospital has offered weekly, multidisciplinary evaluations of children with DS under the age of 3. Serving as a tertiary referral-based clinic, the Program is positioned within the Developmental Medicine Center in the Department of Medicine. Beginning in 2009, the Program expanded its services to include a separate weekly, multidisciplinary evaluation of children with DS between the ages of 3 and adulthood. This new clinic includes a visit with a physician who specializes in Down syndrome (developmental–behavioral pediatrician and/or medical geneticist), a nutritionist, an audiologist, resource specialists, and, when needed and available, other healthcare professionals. Prior to their clinical visit, parents and/or guardians are asked to complete a comprehensive intake questionnaire, which includes information about pre-existing conditions, previous laboratory and radiological studies, and a series of questions about the patient's sleep habits. (Intake available at www.childrenshospital.org/downsyndrome.)

The opening of our new clinic in 2009 afforded an opportunity to ask: What new diagnoses, if any, were identified in our patients with DS as a result of their visit to our clinic? How many of our new patients were up-to-date on the recommended DS healthcare screenings? And, ultimately, what value does a DS specialty clinic bring to people with DS and their families?

MATERIALS AND METHODS

Patients

We analyzed the records of all patients with DS, ages 3 and older, presenting for a new patient visit during the inaugural year of our new clinic (October 3, 2009–October 3, 2010). As we wished to assess the value of our services for children who had not previously had access to a DS clinic, we excluded those patients whom we had seen within the past 3 years in our clinic for younger children. A total of 105 patients were included in our final analyses.

Data Extraction

We reviewed the clinic intake forms and electronic medical records as part of a quality improvement initiative to answer the following questions: (1) Prior to their clinical visit, were patients up-to-date on the healthcare screening recommended by the AAP and DSMIG? During the first year our new clinic was opened, physicians were guided by healthcare recommendations from the AAP [American Academy of Pediatrics. Committee on Genetics, 2001] and the DSMIG [Van Cleve and Cohen, 2006; Van Cleve et al., 2006; Cohen, W.I. for the Down Syndrome Medical Interest Group, 1999] which have since been updated and replaced by newer guidelines from the AAP [Bull and Committee on Genetics, 2011]. (2) What were the most common pre-existing, co-occurring diagnoses in our patients? (3) What were the top concerns of their parents or guardians when coming to our specialty clinic? (4) What laboratory tests, radiology studies, and subspecialty referrals did we order/make as a result of the clinical visit? (5) What new co-occurring diagnoses did we make in our patients as a direct result of their clinical visit? Some of the data for the first three questions were originally obtained from our clinic's intake forms, completed by the parents, and, whenever possible, confirmed by medical records. The data for the last two questions were obtained by reviewing our electronic medical record system. At least one primary researcher coded each patient's record; approximately 16% were coded by a second researcher who was blinded to the first researcher's codes, with coding agreement achieved at 95%. Discrepancies were discussed, and mutual agreement was achieved.

Analysis

Summary statistics (means, standard deviations, and percentages) were used to answer our a priori questions. We report those categories that were represented in at least 4% of the patients. A Fisher exact test was used to assess significance on parentally completed sleep questionnaires between those patients who had obstructive sleep apnea on polysomnograms and those who did not.

RESULTS

Respondents

The 105 patients included in this analysis were, on average, 9.5 years old (SD = 3.8, range = 3.2–20.9). The majority was male (63.8%).

Patients' Status on Healthcare Guidelines

Approximately 78% of our patients were up-to-date on their X-ray screenings for occiptoaxial instability (OAI) and atlantoaxial instability (AAI), but around half of the patients were current on their hearing screens, vision screens, thyroid tests, and celiac disease screen (Table I). Overall, only 9.8% of patients were clinically up-to-date on all of the recommendations from the AAP [American Academy of Pediatrics. Committee on Genetics, 2001] and DSMIG [Van Cleve and Cohen, 2006; Van Cleve et al., 2006; Cohen, W.I. for the Down Syndrome Medical Interest Group, 1999] prior to their appointment in our clinic. If only the recommendations from the AAP were assessed, 16.7% patients were fully up-to-date.

Pre-Existing, Co-Occurring Medical Conditions

Prior to their first clinical appointment in our specialty clinic, our patients had already undergone many previous surgeries, including cardiac repairs (e.g., atrioventricular septal defects, atrial septal defects, ventricular septal defects, and patent ductus ateriosus

TABLE I. Patient's Status per DS Healthcare Guidelines [American Academy of Pediatrics. Committee on Genetics, 2001; Van Cleve and Cohen, 2006; Van Cleve et al., 2006; Cohen, W.I. for the Down Syndrome Medical Interest Group, 1999]

Test	AAP guidelines	DSMIG guidelines	Number patients with complete records	Number up-to-date	% Up-to-date
Thyroid function tests	Annually	Annually	103	58	56.3
Audiograms	Annually	Annually	104	49	47.1
Celiac screen	None	At 2 years	104	47	45.2
Ophthalmology exam	Annually	Annually	104	58	55.8
Cervical spine X-ray	At 3 years	At 3 years	104	81	77.9
All the above			102	10	9.8

TABLE II. Pre-Existing Diagnoses in Patients With Down Syndrome Prior to Clinic Visit (N = 105)*

Diagnosis	Number	%
Cardiac diagnoses		
s/p atrioventricular septal defect repair	24	22.9
s/p ventricular septal defect repair	14	13.3
s/p patent ductus arteriosis	12	11.4
s/p atrial septal defect repair	10	9.5
Otorhinolaryngologic diagnoses		
s/p myringotomy/tympanostomy	56	53.3
s/p adenoid and/or tonsil surgery	54	51.4
Seasonal allergies	19	18.1
Endocrinologic diagnoses		
Hypothyroidism	21	20.0
Ophthalmologic diagnoses		
Strabismus	21	20.0
Hyperopia	19	18.1
Astigmatism	10	9.5
Myopia	9	8.6
Pulmonary diagnoses		
Pneumonia	21	20.0
RSV bronchiolitis hospitalization	14	13.3
Asthma/reactive airway disease	11	10.5
Obstructive sleep apnea	10	9.5
Gastroenterologic diagnoses		
Constipation	16	15.2
Gastroesophageal reflux	14	13.3
Gastroenteritis/dehydration hospitalization	7	7.7
Audiologic diagnoses		
Conductive hearing loss	12	11.4
Hearing loss unspecified	9	8.6
Sensorineural hearing loss	6	5.7
Orthopedic diagnoses		
Ankle instability, hypermobility	8	7.6
Scoliosis	8	7.6
Atlantoaxial instability	6	5.7
Hip problems	6	5.7
Other diagnoses		
Expressive language disorder	11	10.5
Seizures	5	4.8
Autism spectrum disorder	5	4.8
Eczema	5	4.8

*A patient could have more than one diagnosis; as such, percentages do not add up to 100%.

closures) and otolaryngologic procedures (e.g., myringotomies and adenotonsillectomies) (Table II). Their pre-existing, co-occurring medical conditions included ophthalmologic diagnoses (e.g., myopia, hyperopia, astigmatism and strabismus), gastrointestinal issues (e.g., constipation and gastroesophageal reflux disorder), respiratory concerns (e.g., pneumonia, seasonal allergies, RSV bronchiolitis, asthma and obstructive sleep apnea), endocrine conditions (e.g., hypothyroidism), and neurologic concerns (e.g., atlantoaxial instability and seizures).

Top Parental Concerns

Parents and guardians who visit our DS clinic with their children wanted to discuss a range of issues, with the most common questions related to difficult-to-manage behaviors, expressive language challenges, and their child's weight (Table III). Other topics included questions related to orthopedic, dental, gastrointestinal, auditory, and ophthalmologic concerns. Many parents also wanted to talk about concerns related to their child's sleep.

Laboratory Tests, Radiologic Studies, and Referrals

As a result of the clinic visit, we ordered many tests to bring the patients up-to-date in accordance with DS healthcare guidelines

TABLE III. Parental Concerns During Clinical Visit (N = 105)*

Tests	Number	%
Behavior problems	57	54.3
Language concerns	56	53.3
Eating/weight/diet issues	20	19.0
Orthopedic issues	17	16.2
Educational concerns	16	15.2
Sleeping issues	13	12.4
Constipation	10	9.5
Questions about DS guidelines	9	8.6
Potty training	9	8.6
Vision concerns	7	6.7
Dental concerns	6	5.7
Hearing concerns	5	4.8

*A parent could have more than one concern; as such, percentages do not add up to 100%.

that were available at that time (e.g., thyroid tests, celiac screens, audiograms, vision exams and lateral neck radiographs) (Table IV). We often ordered polysomnograms for clinical concerns about obstructive sleep apnea and abdominal X-rays for concerns about constipation. We generally ordered CBCs and/or hip X-rays when there were specific concerns about leg pain or limping, depending on the location and duration of the pain. We offered Fragile X syndrome testing, among other genetic testing, when a dual diagnoses of autism was established, as recommended by the American College of Medical Genetics [Schaefer et al., 2008].

When we appreciated expressive language difficulties, we often made—and parents followed through on—referrals, as appropriate, to the Augmentative Communication Program and Speech And Language Pathology Department at our hospital (Table V). For further diagnostic work-up for difficult-to-manage behaviors, we referred to clinical psychology. When new thyroid-related diagnoses were made, we referred to endocrinology. When a new diagnosis of obstructive sleep apnea was made in a patient who still had tonsils, we referred to otorhinolaryngology. If an adenotonsillectomy had already been performed in these patients, we referred to otorhinolaryngology for adenoidal regrowth evaluation followed by, if needed, a referral to pulmonology/sleep medicine for continuous positive airway pressure (CPAP) evaluations. Of the 15 patients with newly diagnosed obstructive sleep apnea, nine already had an adenotonsillectomy, three followed through with only an otorhinolaryngology referral, three followed through with only a pulmonology referral only, and one followed through with both referrals.

New Co-Occurring Diagnoses

As a direct result of their clinical visits, the patients with DS were diagnosed with many new co-occurring conditions (Table VI). Among these included gastrointestinal conditions (e.g., constipation and celiac disease), seasonal allergies, dermatologic conditions (e.g., xerosis), behavioral diagnoses (e.g., autism spectrum disorder and disruptive behavior NOS), and clarification of neurologic conditions (e.g., AAI). Many patients had hearing loss; many also had an expressive language disorder.

Of the 56 families who had a primary concern about behavioral issues during their initial visit, their sons and daughters were ultimately diagnosed with expressive language disorders, constipation, hearing disorder, ophthalmologic diagnoses, obstructive sleep apnea, thyroid disorders, autism spectrum disorder, and/or celiac disease (Table VII). Our six patients with newly diagnosed autism spectrum disorder ranged in age from 3.3 to 7.7 years old (mean = 6.2 years). When asked on our clinical intake form, 83.5% of all parents felt that their child's Individual Education Plan (IEP) was meeting their educational needs (N = 105). Of those 56 parents

TABLE V. Referrals Completed as a Result of Clinical Visit (N = 105)*

Referral	Number	%
Augmentative communication	43	41.0
Psychology	31	29.5
Speech therapy	27	25.7
Ophthalmology	15	14.3
Pulmonology	9	8.6
Dentistry	7	6.7
Physical therapy	7	6.7
Endocrinology	7	6.7
Occupational therapy	6	5.7

*A patient could have more than one referral; as such, percentages do not add up to 100%.

TABLE IV. Studies/Tests Completed Because of Clinical Visit (N = 105)*

Test/study	Number	%
Thyroid function tests	60	57.1
Celiac screen	55	52.4
Audiogram	46	43.8
Sleep study	43	41.0
Abdominal X-ray	31	29.5
Lateral neck radiograph	27	25.7
CBC	15	14.3
Ophthalmology exam	13	12.4
Basic chemistry panel	10	9.5
Hip X-ray	6	5.7
Fragile X syndrome testing	5	4.8
Acylglycines	5	4.8
MRI, excluding brain MRI	5	4.8

*A patient could have more than one diagnostic test performed; as such, percentages do not add up to 100%.

TABLE VI. New Diagnoses Made as a Result of Clinical Visit (N = 105)*

Diagnosis	Number	%
Xerosis	57	54.3
Expressive language disorder	56	53.3
Disruptive behavior NOS	39	37.1
Obesity (>95% BMI)	27	25.7
Overweight (85–95% BMI)	22	21.0
Constipation	20	19.0
Seasonal allergies	19	18.1
Obstructive sleep apnea	15	14.3
Thyroid conditions[a]	9	8.6
Celiac disease	6	5.7
Autism spectrum disorder	6	5.7
Sensorineural hearing loss	5	4.8
Hearing loss, unspecified	5	4.8
Atlantoaxial instability	5	4.8
Removed atlantoaxial instability diagnosis	5	4.8

*A patient could have more than one diagnosis; as such, percentages do not add up to 100%.
[a]Included one patient with hypothyroidism, four patients with Hashimoto's thyroiditis, and four patients with hyperthyrotropinemia (compensated hypothyroidism).

TABLE VII. New Diagnoses for Patients Whose Parents Had an Initial Concern of Behavioral Issues (N = 56)*

Diagnosis	Number	%
Expressive language disorder	39	72.2
Disruptive behavior disorder NOS	37	66.1
Constipation	19	33.9
Hearing disorder	12	21.4
Ophthalmologic diagnosis	11	19.6
Obstructive sleep apnea	9	16.1
Thyroid disorder	5	8.9
Autism spectrum disorder	5	8.9
Celiac disease	4	7.1

*A patient could have more than one diagnosis; as such, percentages do not add up to 100%.

who had behavioral concerns for their child, 78.6% felt their child's IEP was meeting their needs. Before the clinical encounter, many families were unaware of their child's right to receive behavioral services within the school system and unsure where else in the community to turn for resources.

Parents were further asked on the clinical intake form about symptoms related to obstructive sleep apnea (e.g., "Does your child snore at night?"). Of those patients who went on to have polysomnograms as a result of their clinical visit, there was no statistically significant difference between the responses of parents whose children had obstructive sleep apnea and those who did not. (Sleep apnea questions included on our clinic's intake form are available at www.childrenshospital.org/downsyndrome).

DISCUSSION

The patients with DS, ages 3 and older, who visited our new specialty clinic during its inaugural year had many pre-existing diagnoses, typical for people with DS, but fewer than 10% of them were current on all of the screening guidelines that were in place at the time. Most patients had already had already been screened for OAI and AAI with a cervical spine X-ray, likely because the radiologic test is a requirement for participation in Special Olympics and other athletic programs. For those who had not, several were diagnosed with AAI and placed on activity restriction after an X-ray was performed. A number of patients who previously carried the diagnosis of AAI were released from such restrictions after new X-rays performed in our clinic showed atlanto-dens intervals <4.5 mm and neural canal widths greater than age-specific lower limits [Cohen, 2006]. After our study was complete, the AAP issued new healthcare guidelines, recommending that lateral neck radiographs only be performed when patients with DS had symptoms concerning for OAI or AAI [Bull and Committee on Genetics, 2011]. As the guidelines indicate, "Plain radiographs do not predict well which children are at increased risk of developing spine problems, and normal radiographs do not provide assurance that a child will not develop spine problems later" [Bull and Committee on Genetics, 2011].

About half of the patients did not have ophthalmologic and audiologic evaluations within the past year. This might be secondary to the lack of services in their area, parental choice not to pursue testing, or an oversight in ordering the screens in the primary care setting. After such evaluations were arranged through our clinic, nearly 10% of our patients were diagnosed with sensorineural or unspecified hearing loss. Per the new AAP guidelines, for a child who passes diagnostic hearing testing, "additional screening or behavioral audiogram and tympanometry should be performed every 6 months until normal hearing levels are established bilaterally by ear-specific testing (usually after 4 years of age). Subsequently, behavioral hearing tests should be performed annually" [Bull and Committee on Genetics, 2011]. Further, since our study, the AAP now recommends that ophthalmologic examinations be performed at 6 months of age, annually between the ages of 1 and 5, every 2 years between the ages of 5 and 13, and every 3 years between the ages of 13 and 21 [Bull and Committee on Genetics, 2011].

Approximately 56% of our patients were up-to-date on their annual thyroid screens, in comparison to the 13% of patients with DS in Oklahoma and 14% of patients with DS in Nebraska [Fergeson et al., 2009]. The difference might be explained by our patient samples: our study involved a referred population, whose parents were already motivated to seek tertiary care; the previous study mostly used a population-based sample. Regional variances in primary care practice might also contribute to the difference. Many of our patients were diagnosed with new thyroid conditions, and the AAP continues to recommend that all patients with DS be screened for thyroid conditions on an annual basis [Bull and Committee on Genetics, 2011].

Approximately 45% of our patients had been tested for celiac disease at least once in their lifetime. Lack of services, parental choice not to pursue testing, or an oversight in ordering the screens in the primary care setting might explain, in part, why not everyone received the screening. Additionally or alternatively, primary care physicians might have been following the AAP guidelines [American Academy of Pediatrics. Committee on Genetics, 2001] and not the DSMIG guidelines [Van Cleve and Cohen, 2006; Van Cleve et al., 2006; Cohen, W.I. for the Down Syndrome Medical Interest Group, 1999] which provided discrepant screening recommendations (Table I). Additional research has also questioned the cost effectiveness of screening all asymptomatic patients with DS [Swigonski et al., 2006]. When patients had not yet been tested or when they presented with symptoms concerning for celiac disease, we often ordered blood screens. As a result, about 6% screened positive and were ultimately confirmed to have celiac disease after intestinal biopsies. Since our study, the AAP now recommends that physicians review for symptoms potentially related to celiac disease at every health maintenance visit and evaluate, if indicated, with further testing [Bull and Committee on Genetics, 2011].

Approximately 15% of our patients had a pre-existing diagnosis of chronic constipation, and another 19% received a new diagnosis, underscoring the increased prevalence in our referred population [Chicoine and McGuire, 2010]. Often, our patients or their parents reported troubling symptoms, which included abdominal cramps, bloating, flatus, and concomitant behavioral problems. Fortunately, many of our patients found symptomatic relief after a bowel clean-out regimen performed at home; when this failed, some were admitted to the hospital for a nasogastric tube administration of

GoLytely solution. Constipation in patients with DS can also be secondary to other underlying conditions, including hypothyroidism, celiac disease, and tethered spinal cord, among others, which we evaluated in our clinic.

Many of our patients with DS whose parents were concerned about new disruptive behaviors were diagnosed with medical concerns that might explain, in full or in part, those behaviors: constipation, hearing disorders, ophthalmologic diagnoses, thyroid disorders, obstructive sleep apnea, and/or celiac disease. New behavioral problems are sometimes the only indications that children and adolescents with DS are feeling discomfort or that there are changes in their health [McGuire and Chicoine, 2006]. For some patients, their disruptive behaviors were secondary to expressive language frustrations, which could be mitigated, to varying degrees, with the assistance of augmentative communication devices and additional speech and language therapy.

In some cases, after a clinical psychologist at our hospital performed a full neuropsychological assessment, a dual diagnosis of autism spectrum disorder was made, which has been appreciated to occur at a higher incidence in children with DS [Ji et al., 2011; McGrath et al., 2011]. Our findings were also consistent with previous studies which demonstrate a later age of diagnosis of autism in children with DS compared with the general population, highlighting the need for screening in this population [DiGuiseppi et al., 2010]. As a result of their new diagnosis of autism spectrum disorder, many of our patients began to receive Applied Behavioral Analysis (ABA) therapy and were given resources for community supports.

For many of our patients whose parents had behavioral concerns, a secondary cause or diagnosis was not identified, and the patients were given a de facto diagnosis of Disruptive Behavior Disorder, Not Otherwise Specified. In these circumstances, many of the parents requested specific guidance on behavioral interventions and management options, both for the home and school settings. We oftentimes worked closely with a pediatric psychologist at our hospital, who developed a booklet and a Webinar for parents, in addition to meeting them in individual and group sessions. That so many parents listed disruptive behaviors as a concern during our clinic visit underscores the need for DS specialty clinics to include or collaborate with pediatric psychologists.

Many of our patients also met the clinical definition of being overweight or obese, which speaks to the importance of having a nutritionist be part of the regular care of persons with DS. As obesity can lead to secondary conditions such as diabetes, to which people with DS are already predisposed, establishing and maintaining healthy lifestyles during childhood is important [Medlen, 2012]. Each patient in our multidisciplinary clinic is offered an opportunity to meet with a nutritionist. The latest AAP guidelines recommend that since ''previously used Down syndrome specific growth charts no longer reflect the current population styles and body proportion,'' all children and adolescents with DS should be evaluated by body mass index (BMI) or weight-for-height trends on standard growth curves from the National Center for Health Statistics or the World Health Organization [Bull and Committee on Genetics, 2011].

Approximately 14% of our patients were newly diagnosed with obstructive sleep apnea, but not all families followed through with our referrals to see an otorhinolaryngology and/or sleep medicine physician. This could be due to parental preference or the possibility that some families sought referral care outside of our hospital system, in which those clinical visits would not be captured in our electronic medical records. Our clinical intake form asked parents 13 questions about symptoms that are concerning for obstructive sleep apnea, a condition associated with significant morbidity including short- and long-term cognitive deficits [Halbower et al., 2006], behavioral disturbances [Mitchell and Kelly, 2007], attention deficit and hyperactivity disorder [Huang et al., 2007], school failure [Brouillette et al., 1982], hypertension [Li et al., 2008], poor glucose metabolism [Tamura et al., 2008], increased cardiovascular and cerebrovascular disease [Parish and Somers, 2004; Nishibayashi et al., 2008], failure to thrive [Brouillette et al., 1982], pulmonary hypertension [Brouillette et al., 1982], and even death [Bradley and Phillipson, 1985]. Compared to the typical pediatric population, where only 1–4% of children are estimated to have obstructive sleep apnea [Lumeng and Chervin, 2008], the prevalence in patients with Down syndrome is very high with estimates ranging between 55% and 97%, although many of these studies used referred symptomatic populations [Marcus et al., 1991; de Miguel-Diez et al., 2003; Dyken et al., 2003; Ng et al., 2006; Shott et al., 2006; Fitzgerald et al., 2007]. Our results showed that parents whose children had obstructive sleep apnea did not respond any differently on these questions than parents whose children did not have obstructive sleep apnea. This finding is consistent with a previous study and underscores the challenge in using parental concerns, alone, to further investigate obstructive sleep apnea [Shott et al., 2006]. To this extent, the latest AAP guidelines recommend that all children with DS, even those asymptomatic, have a sleep study performed by the age of four [Bull and Committee on Genetics, 2011].

Beyond the medical care patients received in our clinic, families also received updates on community-based resources and supports. A Resource Specialist met with each family and referred them to local conferences, social functions, books, and support organizations, among other resources. A person with DS also worked in our clinic and was able to direct families to many of these supports, while also sharing his personal life experiences [Coldwell, 2012]. Many families who visit DS specialty clinics across the US benefit from the staff's knowledge about the latest information on clinical, research, and advocacy efforts. Oftentimes, this information is shared with families during clinic visits, e-newsletters, web pages, and Webinars [Children's Hospital Boston, 2012].

Our quality improvement study had several limitations, including selection bias. Our patients come from a referred population, and do not necessarily represent all families who have children and adolescents with DS. Families whose sons and daughters had more complex medical needs might have been more likely to seek our consultation, especially during our inaugural year. As such, our percentages of pre-existing and newly made diagnoses do not necessarily represent the incidence of these co-occurring conditions in all people with DS. Nonetheless, the multiple parental concerns and varied diagnostic evaluations underscore the unmet needs of many patients with DS in our area. Our study is also limited by the boundaries of a retrospective electronic chart review. In many cases, we had clinical concerns, prompting referrals to subspeciality services, but if the parents did not follow-up on those recommen-

dations or sought consultation outside of our hospital system, some newly made diagnoses might not have been identified in our electronic medical system. As such, the percentages of our newly made diagnoses represent, if anything, a lower limit for our patient sample. Additionally, our study is subject to some parental recall bias. Whenever possible, we attempted to confirm through medical records all previous laboratory results and pre-existing diagnoses. When not possible or practical, however, we relied on parental reporting, as is customary for clinical practice. To this extent, we could not exclude parents who knowingly or unknowingly misrepresented the truth.

Unanswered in our study are the reasons why parents sought out our DS specialty clinic in the first place. Future prospective studies should further ask parents: What type of primary care have they established for their son or daughter? What healthcare recommendations had they received from their child's provider? What is the family's awareness of the revised DS healthcare guidelines from the AAP? Is their son or daughter up-to-date on other routine aspects of medical care, such as immunizations and sexuality counseling? Determining the reasons that patients were not current on specific guidelines could facilitate interventions to improve compliance and optimal care within the medical home.

While this qualitative study did capture benefits that our clinic provided to our patients and their families, the study is by no means an exhaustive assessment of such value. Further studies can and should tackle the implications of our interventions: Did the patients respond to new treatments? Were there reduced morbidity and mortality after a new diagnosis was made? Did the behavior problems improve after underlying diagnoses were identified and treated? The establishment of a national Down syndrome patient database would greatly assist efforts in answering these questions.

Without exception, the patients seen in our clinic still need quality primary care. We saw most patients once a year and did not typically serve their routine pediatric needs. Healthcare for children and adolescents with DS can be improved by systematic use of the new AAP guidelines [Bull and Committee on Genetics, 2011] and the establishment of a medical home in the primary care setting [Council on Clinical Information Technology, 2011]. Online courses with continuing medical education credits have been created for primary care physicians seeking to learn more about managing the healthcare of patients with DS [Skotko, 2009]. Additionally, the National Down Syndrome Society has created age-specific checklists, based on the new AAP guidelines, that can be accessed by parents and providers (http://www.ndss.org/Resources/Health-Care/Health-Care-Guidelines/).

However, our research suggests that the increasing complexities of DS screening might demand time and attention beyond that which is possible for today's primary care providers. According to one nationally representative study, only 29.7% of children with DS have an established medical home and are more than two times more likely to have unmet needs for care and family support than children with other special healthcare needs [McGrath et al., 2011]. This challenge is not unique to the United States; in Finland, researchers found that the health surveillance of people with DS was "insufficient," as well [Maatta et al., 2011]. Of course, with a growing number of guidelines being developed for many genetic and developmental conditions, this challenge might also not be unique to DS. For example, one study found that more than 50% of patients with Turner syndrome treated at Riley Hospital in Indianapolis, IN, were "deficient" in their recommended screening for associated co-occurring conditions [Nabhan and Eugster, 2011].

To this extent, DS specialty clinics can serve as important collaborators in providing the comprehensive medical care that is needed. These clinics can be especially helpful for the evaluation of challenging co-occurring diagnoses, especially disruptive behavior disorders, autism spectrum disorder, and expressive language problems. National Down syndrome organizations are currently advocating for the passage of the Trisomy 21 Research Centers of Excellence Act of 2011, which would create more DS clinics to provide optimal venues for clinical care and translational research [National Down Syndrome Society, 2012b]. Our research demonstrates that such funding would serve an important and functional need in maximizing the wellness of children and adolescents with DS.

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REFERENCES

American Academy of Pediatrics. Committee on Genetics. 2001. Health supervision for children with Down syndrome. Pediatrics 107:442–449.

Bradley TD, Phillipson EA. 1985. Pathogenesis and pathophysiology of the obstructive sleep apnea syndrome. Med Clin North Am 69:1169–1185.

Brouillette RT, Fernbach SK, Hunt CE. 1982. Obstructive sleep apnea in infants and children. J Pediatr 100:31–40.

Bull MJ, Committee on Genetics. 2011. Health supervision for children with Down syndrome. Pediatrics 128:393–406.

Chicoine B, McGuire D. 2010. The guide to good health for teens & adults with Down syndrome. Bethesda: Woodbine House.

Children's Hospital Boston. 2012. Speaker series presentations. Available at http://www.childrenshospital.org/clinicalservices/Site2845/mainpageS2845P5.html. Accessed on November 3, 2012.

Cohen WI, for the Down Syndrome Medical Interest Group. 1999. Health care guidelines for individuals with Down syndrome: 1999 revision. Downs Syndr Q 4:1–16.

Cohen WI. 2006. Current dilemmas in Down syndrome clinical care: Celiac disease, thyroid disorders, and atlanto-axial instability. Am J Med Genet Part C 142C:141–148.

Coldwell S. Children's new star player. Available at http://www.childrenshospital.org/dream/winter10/childrens_new_star_player_winter10.html. Acccessed on November 3, 2012.

Council on Clinical Information Technology. 2011. Health information technology and the medical home. Pediatrics 127:978–982.

de Miguel-Diez J, Villa-Asensi JR, Alvarez-Sala JL. 2003. Prevalence of sleep-disordered breathing in children with Down syndrome: Polygraphic findings in 108 children. Sleep 26:1006–1009.

DiGuiseppi C, Hepburn S, Davis JM, Fidler DJ, Hartway S, Lee NR, Miller L, Ruttenber M, Robinson C. 2010. Screening for autism spectrum disorders in children with Down syndrome: Population prevalence and screening test characteristics. J Dev Behav Pediatr 31:181–191.

Dyken ME, Lin-Dyken DC, Poulton S, Zimmerman MB, Sedars E. 2003. Prospective polysomnographic analysis of obstructive sleep apnea in Down syndrome. Arch Pediatr Adolesc Med 157:655–660.

Fergeson MA, Mulvihill JJ, Schaefer GB, Dehaai KA, Piatt J, Combs K, Bright BC, Neas BR. 2009. Low adherence to national guidelines for thyroid screening in Down syndrome. Genet Med 11:548–551.

Fitzgerald DA, Paul A, Richmond C. 2007. Severity of obstructive apnoea in children with Down syndrome who snore. Arch Dis Child 92:423–425.

Halbower AC, Degaonkar M, Barker PB, Earley CJ, Marcus CL, Smith PL, Prahme MC, Mahone EM. 2006. Childhood obstructive sleep apnea associates with neuropsychological deficits and neuronal brain injury. PLoS Med 3:e301.

Huang YS, Guilleminault C, Li HY, Yang CM, Wu YY, Chen NH. 2007. Attention-deficit/hyperactivity disorder with obstructive sleep apnea: A treatment outcome study. Sleep Med 8:18–30.

Jensen KM, Taylor LC, Davis MM. 2012. Primary care for adults with Down syndrome: Adherence to preventive healthcare recommendations. J Intellect Disabil Res DOI: 10.1111/j.1365-2788.2012.01545.x. [Epub ahead of print].

Ji NY, Capone GT, Kaufmann WE. 2011. Autism spectrum disorder in Down syndrome: Cluster analysis of aberrant behaviour checklist data supports diagnosis. J Intellect Disabil Res 55:1064–1077.

Li AM, Au CT, Sung RY, Ho C, Ng PC, Fok TF, Wing YK. 2008. Ambulatory blood pressure in children with obstructive sleep apnoea: A community based study. Thorax 63:803–809.

Lumeng JC, Chervin RD. 2008. Epidemiology of pediatric obstructive sleep apnea. Proc Am Thorac Soc 5:242–252.

Maatta T, Maatta J, Tervo-Maatta T, Taanila A, Kaski M, Iivanainen M. 2011. Healthcare and guidelines: A population-based survey of recorded medical problems and health surveillance for people with Down syndrome. J Intellect Dev Disabil 36:118–126.

Marcus CL, Keens TG, Bautista DB, von Pechmann WS, Ward SL. 1991. Obstructive sleep apnea in children with Down syndrome. Pediatrics 88:132–139.

McGrath RJ, Stransky ML, Cooley WC, Moeschler JB. 2011. National profile of children with Down syndrome: Disease burden, access to care, and family impact. J Pediatr 159:535.e2–540.e2.

McGuire D, Chicoine B. 2006. Mental wellness in adults with Down syndrome: A guide to emotional and behavioral strengths and challenges. Bethesda, MD: Woodbine House.

Medlen J. 2012. Down syndrome nutrition. Available at http://downsyndromenutrition.com/. Accessed on November 3, 2012.

Mitchell RB, Kelly J. 2007. Behavioral changes in children with mild sleep-disordered breathing or obstructive sleep apnea after adenotonsillectomy. Laryngoscope 117:1685–1688.

Nabhan ZM, Eugster EA. 2011. Medical care of girls with turner syndrome: Where are we lacking? Endocr Pract 17:747–752.

National Down Syndrome Society. 2012a. Down syndrome clinics and health care providers database. Available at http://www.ndss.org/Resources/Health-Care/Health-Care-Providers/. Accessed on November 3, 2012.

National Down Syndrome Society. 2012b. Legislation and initiatives—Improving health outcomes and quality of life for people with Down syndrome. Available at http://www.ndss.org/Advocacy/Legislative-Agenda/Improving-Health-Outcomes-Quality-of-Life-for-People-with-Down-Syndrome/. Accessed on November 3, 2012.

Ng DK, Hui HN, Chan CH, Kwok KL, Chow PY, Cheung JM, Leung SY. 2006. Obstructive sleep apnoea in children with Down syndrome. Singapore Med J 47:774–779.

Nishibayashi M, Miyamoto M, Miyamoto T, Suzuki K, Hirata K. 2008. Correlation between severity of obstructive sleep apnea and prevalence of silent cerebrovascular lesions. J Clin Sleep Med 4:242–247.

Parish JM, Somers VK. 2004. Obstructive sleep apnea and cardiovascular disease. Mayo Clin Proc 79:1036–1046.

Presson AP, Partyka G, Jensen KM, Rasmussen SA, Devine OJ, McCabe L, McCabe ERB. 2012. Prevalence of Down syndrome in the United States. Presented at American Society for Human Genetics, 2012, San Francisco.

Schaefer GB, Mendelsohn NJ, Professional Practice and Guidelines Committee. 2008. Clinical genetics evaluation in identifying the etiology of autism spectrum disorders. Genet Med 10:301–305.

Shott SR, Amin R, Chini B, Heubi C, Hotze S, Akers R. 2006. Obstructive sleep apnea: Should all children with Down syndrome be tested? Arch Otolaryngol Head Neck Surg 132:432–436.

Skotko BS. 2009. Down syndrome: Healthcare updates for the primary care pediatrician. Available at http://cmeonline.med.harvard.edu/course_descriptions.asp?Course_id=121. Accessed on November 3, 2012.

Swigonski NL, Kuhlenschmidt HL, Bull MJ, Corkins MR, Downs SM. 2006. Screening for celiac disease in asymptomatic children with Down syndrome: Cost-effectiveness of preventing lymphoma. Pediatrics 118:594–602.

Tamura A, Kawano Y, Watanabe T, Kadota J. 2008. Relationship between the severity of obstructive sleep apnea and impaired glucose metabolism in patients with obstructive sleep apnea. Respir Med 102:1412–1416.

Van Cleve SN, Cohen WI. 2006. Part I: Clinical practice guidelines for children with Down syndrome from birth to 12 years. J Pediatr Health Care 20:47–54.

Van Cleve SN, Cannon S, Cohen WI. 2006. Part II: Clinical practice guidelines for adolescents and young adults with Down syndrome: 12–21 years. J Pediatr Health Care 20:198–205.