

A Community-Health System Intervention to Improve the Primary Healthcare of Adults with
Down Syndrome Through Electronic Consultations

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Abstract

Health systems often fail to tap the expertise of the developmental disabilities community support and service system. In this 9-month pilot health system-level quality improvement project, a multi-disciplinary team of physician, pharmacist, and disabilities advocate reviewed electronic records of patients with Down syndrome in advance of pre-scheduled appointments with their primary care physician and generated 100 electronic consultations. Post-consultation chart review documented meaningful uptake of clinical recommendations, including screening for thyroid disease, celiac disease and heart disease, pneumococcal vaccination, and screening physical examination for myelopathy. In addition to clinical recommendations regarding screening, diagnosis, and treatment, each consultation provided an average of 8 tailored suggestions for potential community-based resources related to mental and behavioral health, recreation, socialization, and other relevant services and supports. “Push” multi-disciplinary electronic consultations in advance of primary care appointments enriched with input from disabilities community experts have the potential to improve the quality of health care provided to persons with developmental disabilities.

Key Words: *electronic consultation; clinical decision supports; Down syndrome; health system-community partnership; health system- quality of service*

Introduction

Assuring high-quality, tailored healthcare for persons with low-prevalence or rare conditions is an intrinsic challenge to primary care. As an example, Down syndrome (DS), the most common chromosomal cause of intellectual disabilities, occurs at a rate of 1:691 births.¹ While many primary care physicians are responsible for one or two adults with Down syndrome in their clinic panels, they may be unfamiliar with the healthcare needs specific to DS.² In their analysis of electronic health record data examining health services provided adults with Down syndrome, Jensen et al. documented that less than 50% of patients with DS were screened for common co-morbid conditions such as obstructive sleep apnea, atlanto-axial instability, hearing loss and vision loss.³ In addition to a lack of knowledge about specific medical and other co-morbidities and individualized screening requirements for persons with low prevalence conditions, primary care physicians may be unacquainted with valuable local support, advocacy and service systems and resources relevant to those individuals and their caregivers.

Clinical decision supports embedded in electronic health record systems such as practice alerts, checklists and templates offer one method for improving the healthcare of persons with uncommon conditions with which primary care physicians have little clinical familiarity.¹ However, there is a limit to the number of such electronic supports that health systems can reasonably create or that physicians can realistically respond to.⁴ Additionally, these clinical supports often fall short in failing to provide individualized recommendations for community-based resources and referrals.

This quality improvement pilot project examined the feasibility and preliminary outcomes of a health system-level intervention to improve the primary care of adults with DS by instituting a multi-disciplinary electronic consultation team. Electronic consultations have been defined as “asynchronous, consultative, provider-to-provider communications within a shared

electronic health record or web-based platform.”⁵ Primary aims of this report were to: (1) describe the clinical support intervention; (2) summarize selected content and uptake of Electronic Consultation Team clinical recommendations; and (3) determine whether the primary care physician recipients of these electronic consultations found them valuable.

Methods

This was a quality improvement project designed to examine the feasibility and impact of a multi-disciplinary Electronic Consultation Team (e-CT) on the quality of primary care provided to adults with Down syndrome. The target of this intervention was a cohort of Cleveland Clinic primary care physicians (approximately 200 general internists and family physicians) providing care to adults with Down syndrome through pre-scheduled appointments during the 9-month study period.

Project Protocol

This pilot quality improvement project was approved by the Institutional Review Board of the Cleveland Clinic under expedited review and was funded through a \$50,000 grant from Special Hope Foundation. Utilizing an electronic medical record-based algorithm previously developed by Jensen et al., a list of all adults with Down syndrome age 18 and older who were treated within the prior three years by a primary care physician within the Cleveland Clinic Health system in northeastern Ohio was generated.⁶ On a weekly basis, a list of adults with Down syndrome who were scheduled to see a primary care physician within the following 10 days was provided to the e-CT for chart review and recommendations.

Collaborating community partners who participated as consultants in the project design, interim and final data analysis, interpretation of study findings, and community dissemination of

the project findings were the Developmental Disabilities- Practice-Based Research Network (DD-PBRN) and Up Side of Downs of Northeast Ohio. The DD-PBRN is a community-based, multi-stakeholder group of self-advocates with developmental disabilities, family members of persons with developmental disabilities, residential service providers, health professionals and researchers who conduct research that aims to improve the health and health care of persons with developmental disabilities.⁷ The Up Side of Downs of Northeast Ohio is a local education, support and advocacy organization serving persons with Down syndrome and their families; the project team met with several dyads of adults with Down syndrome and their parents recruited by Up Side of Downs who served as project consultants.⁸

The e-CT was comprised of a family physician with clinical and research expertise in developmental disabilities medicine, a clinical pharmacist, and a representative of the disabilities advocacy community who was an executive director of a local chapter of The Arc.⁹ The e-CT developed a protocol for their consultation process, including a structured electronic medical record (EMR) chart review by the physician and pharmacist, followed by a full team discussion, and generation of clinical recommendations. The physician member focused on recommendations that supported adherence to guidelines for Down syndrome-specific health care, optimum chronic disease management, and evaluation of previously documented signs and symptoms possibly indicative of undiagnosed disease. The clinical pharmacist completed a comprehensive medication review to identify drug- related problems, assuring that medication therapy was appropriate, necessary and optimized, with the goal of identifying sources of poly-pharmacy and reducing pill burden. Additional pharmacotherapy review goals included assessing medication adherence, drug interactions, and opportunities for medication- related cost-savings. Per protocol, as advised by our project community consultants, the disabilities

advocate on the e-CT did not directly examine electronic medical records; however, based on the team discussion, the disabilities advocate recommended potential resources for the patient and caregiver, including residential, education and training ideas, community and social service agencies, and recreational, socialization and volunteer resources. Many of these were drawn from the document *Transition Directory of Cuyahoga County 2015*,¹⁰ along with the disabilities advocate's extensive personal knowledge of local resources relevant to the developmental disabilities community. The e-CT met once weekly for a duration of 2-3 hours, which varied according to the number and complexity of the patients under review.

Following EMR review and team discussion, the physician e-CT member generated a written consultation to the primary care physician (PCP), outlining a list of suggested screening and diagnostic tests, suggestions for further inquiry through history and physical examination, ideas regarding pharmacotherapy, and recommendations regarding referrals, including health care and community-based resources. These consultations were uploaded as an abstract encounter as is typically found in the EMR, accompanied by an email notification alert to the primary care physician, usually several days in advance of the scheduled appointment.

Within one week following the clinical encounter with the adult with Down syndrome, a research assistant contacted the PCP recipient of the electronic consultation to arrange a telephone semi-structured interview. In this interview, the research assistant solicited PCP feedback and critique regarding the structure, content, and delivery of the virtual consultation. The research assistant also administered the Information Assessment Method Cognitive Checklist, an 11-item validated checklist for systematically assessing the quality and impact of email alerts.¹¹

Data Collection

Primary data sources included the text of the virtual consultations; EHR routing data (to determine whether or not the virtual consultation emails were reviewed by the PCP); progress note texts; tests, referrals and medication changes ordered by the PCP; and virtual consultation recipient feedback.

Data Analysis

The clinical recommendations contained within the virtual consultations were organized thematically and tabulated. EHR routing data was examined to determine whether or not the virtual consultation emails were opened by the PCP in advance of the pre-scheduled appointment. The e-CT clinical recommendations were paired against the PCP's documented orders and test results to determine the PCP's implementation and patient uptake of each clinical recommendation. For the purposes of this manuscript, we limited our report to PCP uptake of selected clinical recommendations, a summary of the content of community-based resources and referrals, and physician feedback through the Information Assessment Method Cognitive Checklist. The rationale for the clinical recommendations relate to the increased prevalence of thyroid disease,¹² celiac disease,¹³ acquired valvular heart disease,¹⁴ and invasive pneumococcal disease¹⁵ observed in persons with Down syndrome.

Results

Using the referenced EMR-based algorithm, just over 1100 adults with Down syndrome receiving primary care through the Cleveland Clinic Health System in northeastern Ohio were identified. One-hundred e-consults were conducted over the course of 9 months and 18 post-consultation telephone interviews of physician e-consult recipients were completed. During that

time period, we did not track patients who failed to show for their appointment or patients who were potentially eligible for consultations, yet did not receive them due to factors such as e-CT member scheduling conflicts. Of the weekly lists of adults with Down syndrome with pre-scheduled appointments with a primary care physician, less than 5% of charts reviewed erroneously included persons who clearly did not have Down syndrome; nearly all were women who had undergone prenatal screening for the condition. All of the e-CT recommendations were reviewed by the PCP in advance of the scheduled appointments; this was confirmed by a time-stamped verification within the EMR, which is standard functionality within Epic EMR systems.

Table 1 summarizes demographic characteristics of the patient-subjects for the e-CTs. Most subjects were between ages 20 and 60, with fairly equal distribution each decade within that range. Just over two-thirds of subjects lived with family.

Table 2 summarizes the uptake of selected clinical recommendations encompassing domains of laboratory testing, echocardiography, and immunizations. Because post-consultation chart reviews were conducted within one month of the actual appointments with the primary care physician, tests requiring advanced scheduling, such as echocardiography, may have been ordered, but not yet performed. This is denoted by the column “potential adherence” to clinical recommendation. There was marked variation in the documented uptake of clinical recommendations, from as little as 2% increase in documented referral for dental care, to as much as 25% increase in adherence to screening for thyroid disease by blood test and 26% increase in pneumococcal vaccination rates. Post e-consultation adherence rates tended to be highest for those clinical recommendations with the highest baseline adherence.

On average, each e-consultation offered eight potentially beneficial community-based resources. These suggested resources, primarily the contribution of the disabilities advocate to the e-consultation process, are summarized categorically in Table 3. Nearly all consultations included recommendations to contact Up Side of Downs, the local Down syndrome support and advocacy organization (95%), to investigate a web-based resource for future planning (93%), and to read two core books regarding common medical and mental health issues in adults with Down syndrome (90%). Half of consultations included recommendation to contact The Arc of Greater Cleveland as some type of advocacy, support or referral need was implied by reviewing the medical record (52%). Similarly, nearly half of consultations suggested the clinician review a clinical practice guideline related to evaluation of possible dementia in persons with intellectual and other developmental disabilities (46%).

Table 4 tabulates electronic consultation recipient responses to the Information Assessment Method Cognitive Checklist. Physician responses indicate high satisfaction with the consultations and suggest they enhanced their sense of self-efficacy in providing health care to this population. Qualitative comments from e-CT recipients included: “Lots of meat on the bone;” “Very comprehensive. Hardly see anyone with Down Syndrome. Welcome change. Quick good guidance. Highly recommend to Cleveland Clinic;” “Initially overwhelming in amount; information useful; glad I didn’t ignore the consultation;” “Refresher – helpful. Filled in some gaps and helped the family. Family really appreciated from a medical standpoint and the resources suggested.”

Discussion

This pilot quality improvement project successfully created a partnership of health care institution and community-based expertise to implement a unique health-system level clinical support intervention aiming to address deficiencies in the primary care of adults with Down syndrome. Post-consultation chart review documented meaningful uptake of clinical recommendations. Physicians believed that the electronic consultations improved both their quality of health care and their self-efficacy in providing that care.

Specific uptake of recommendations included a 25% uptake in ordering thyroid stimulating hormone (TSH), a highly sensitive test for thyroid disease, which occurs in up to one-third of individuals with Down syndrome. This diagnosis is easily missed when clinicians rely on clinical features alone in persons with Down syndrome, who often misattribute symptoms of thyroid disease such as obesity, constipation, and dry skin to Down syndrome itself. Consultations yielded a 19% uptake in tissue transglutaminase antibody (TTG-IgA) screening for celiac disease. Celiac disease, also more prevalent in individuals with Down syndrome, is also easily missed without serologic screening in individuals who cannot express symptoms of gastrointestinal distress and are unable to relate this to ingestion of gluten-containing foods.

Strengths of this intervention included efficient utilization of existing health system resources, particularly clinical expertise in the primary care of adults with developmental disabilities, strong electronic record-based research/information technology support, and a mature electronic record system. Equally important for both project design and implementation, community-based expertise was employed, leading to more comprehensive consultations that matched common quality of life issues with practical community resources and information. Because the e-consultations were generated for all adults with Down syndrome scheduled to see a primary care physician, they were not dependent on schedule review in advance of

appointment and pre-emptive request by the primary care physician; in effect, these were “push” consultations based on patient characteristics rather than “pull” consultations dependent upon clinician request. Thus, the clinical support service was not dependent on subjective self-assessments by physicians about their clinical knowledge and resource familiarity related to Down syndrome. This aspect holds broader applicability to other conditions in which clinicians may over-estimate their pragmatic knowledge about relevant community-based resources, supports and services.

Limitations relevant to this project primarily related to the scope of data collection. Since chart review was conducted just once, within weeks of clinic appointment, ultimate uptake of clinical recommendations was likely under-estimated. While we captured physician feedback to the electronic consultation service, there were insufficient resources to allow similar ascertainment of patient or caregiver views of the electronic consultation service. Of note for future work, some of the physicians provided patients a print copy of the entire electronic consultation, allowing both patient and caregiver to see and determine their perceived value of this service.

The clinical members of the e-CT focused on improving adherence to syndrome-specific health screenings, investigating potential undetected disease, and improving the quality of pharmacotherapy. In future iterations of this clinical support service, the quality and scope of e-consultations would be further enhanced by addition of a developmental disabilities nurse to the consultation team, who could provide pragmatic suggestions regarding implementation of clinical recommendations (e.g., cancer screening), disease self-management, and caregiver education. Similarly, including a mental/behavioral health specialist on the team could also

provide invaluable recommendations related to screening, diagnosis and management of psychiatric conditions and behavioral issues.

In an attempt to address gaps in the health care of adults with DS, just over 75 Down syndrome specialty clinics have been established around the United States.¹⁶ While some of these clinics are staffed by multi-disciplinary teams that provide elegant comprehensive evaluation, others provide more limited services. In general, these specialty clinics are difficult to establish and costly to maintain. Electronic consultation models are one means to more broadly improve health care quality at a potentially lesser cost, empower primary care physicians with information and resources to personally deliver more comprehensive care, and thereby efficiently utilize existing, locally accessible health care systems and infrastructure. As more individuals are cared through accountable care organizations, health systems are more willing to fund innovative programs that utilize electronic consultation and care coordination models that foster clinical-community linkages.¹⁸

The Up Side of Downs of Northeast Ohio One was a valued local community consultant to this project. Health professionals are often unaware of the number, extent and reach of such organizations providing education, support, resources and advocacy to persons with specific conditions. The umbrella organization Down Syndrome Affiliates in Action includes over 80 member organizations in the US alone, while the National Down Syndrome Society has identified over 375 Down syndrome organizations worldwide, with the majority in the US.¹⁶ Health systems seeking to meaningfully improve the health care of persons with Down syndrome need to develop relationships with their local and regional Down syndrome support and advocacy organizations.

At the time of this project, the disabilities advocate on the team was director of The Arc of Greater Cleveland. The Arc is the largest developmental disabilities advocacy organization in the United States, with 39 states and 730 local chapters, providing support, education, and advocacy to persons with developmental disabilities and their families.¹⁷ Health systems interested in replicating and expanding this model to serve persons with developmental disabilities are encouraged to contact local chapters of The Arc to similarly develop the health system-community partnerships that we believe were essential to the success of this clinical support service.

We believe this model holds promise for improving the primary care of other developmental disabilities, and more broadly to other low prevalence/rare conditions. This model holds promise for even common conditions and diseases in which an active community-based support and service system exists for patients, but without robust integration with the health system. We encourage community-based organizations and health systems to engage in similar collaborations to develop innovative programs that address health and health care disparities in persons requiring specialized clinical, support and service needs.

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Table 1
Demographic Characteristics of Patient-Subjects of Electronic Consultations (N=100)

| Characteristic | | |
|----------------|-------------|----------|
| Gender | Female | 45 (45%) |
| Age (years) | 18-19 | 1 (1%) |
| | 20-29 | 22 (22%) |
| | 30-39 | 16 (16%) |
| | 40-49 | 19 (19%) |
| | 50-59 | 24 (24%) |
| | 60-69 | 7 (7%) |
| Residence | Family | 69 (69%) |
| | Group | 26 (26%) |
| | Independent | 1 (1%) |
| | Unknown | 4 (4%) |

Table 2
Physician Uptake of Selected e- Consultation Team Clinical Recommendations

| Laboratory | Pre-Consult Adherence | Post-Consult Adherence Immediate / Potential |
|------------|-----------------------|---|
| IgA | 20 (20%) | 39 (39%) / 46 (46%) |
| TTG-IgA | 12 (12%) | 31 (31%) / 40 (40%) |
| TSH | 50 (50%) | 75 (75%) / 81 (81%) |

| Imaging | Pre-Consult Adherence | Post-Consult Adherence Immediate / Potential |
|---------|-----------------------|---|
| Echo | 43 (43%) | 51 (51%) / 61 (61%) |

| Immunizations | Pre-Consult Adherence | Post-Consult Adherence Immediate/Potential |
|---------------|-----------------------|---|
| PCV-13 | 7 (7%) | 33 (33%) / 35 (35%) |
| PVX-23 | 24 (24%) | 28 (28%) / 28 (28%) |

| Physical Exam | Pre-Consult Adherence | Post-Consult Adherence |
|-------------------|-----------------------|------------------------|
| Myelopathy Screen | 6 (6%) | 16 (16%) |
| Testicular Exam | 4/55 (7%) | 13/55 (24%) |
| Otoscopic Exam | 67 (67%) | 89 (89%) |

Table 1
Continued

| Referral | Pre-Consult Adherence | | Post-Consult Adherence | |
|-----------------|-----------------------|-------|------------------------|-------|
| Dentist | 22 | (22%) | 25 | (25%) |
| Ophthalmologist | 27 | (27%) | 31 | (31%) |
| ENT/Audiologist | 16 | (16%) | 28 | (28%) |

Note. “Post-consultation Adherence – Immediate” column refers to tests and services that physician ordered and immediately performed during the index ambulatory encounter; “Post-consultation Adherence – Potential” column refers to all tests and services that physician ordered, whether or not they were immediately performed during the index ambulatory encounter; ENT=Otolaryngologist; IgA=Immunoglobulin A; PCV13= Pneumococcal vaccine 13; PVX 23 = Pneumovax 23; TSH= Thyroid Stimulating Hormone; TTG-IgA= Tissue transglutaminase-Immunoglobulin A

Table 3

Suggested Community-based Resources and Associated Frequencies of Recommendation (N=100)

Community Resources & Frequencies (%)

Up Side of Downs of Northeast Ohio (95); Future Planning (93); The Arc of Greater Cleveland (52); Healthy Lifestyles (28); Ohio Sibling Network (23); Kiwanis Aktion Club (8); Transition-Age Guide (6); Milestones Autism Resources (3); State-support Team Region 8 (2); Dads Appreciating Down Syndrome (D.A.D.S.) (1)

Books & Frequencies (%)

Guide to Good Health for Teens & Adults with Down Syndrome (90); Mental Wellness in Adults with Down Syndrome (90);

Physical Activity/Recreational Resources & Frequencies (%)

Aquatic Exercise (23); Special Olympics (23); SOAR! (17); Best Buddies (17); Socialization through the Arc of Greater Cleveland (14); Silver Sneakers (3); College for Living (2); Mind-Body-Soul (2)

Mental Health Resources & Frequencies (%)

Dementia Clinical Practice Guidelines (46)

Behavioral Resources & Frequencies (%)

County Board of Developmental Disabilities (11)

Other Resources & Frequencies (%)

Cleveland Clinic Home Health Care Services (1); Specs4us (1)

Note. Italics used to indicate book

Table 4

Electronic-Consult Recipient Responses to Information Assessment Method Cognitive Checklist (n=18)

| Item | Yes | No | Maybe |
|---|------|------|-------|
| Through the e-consultation, my practice in caring for persons with Down syndrome was improved. | 94% | 6% | |
| Through the e-consultation, I learned something new about caring for persons with Down syndrome. | 100% | | |
| Through the e-consultation, I am motivated to learn more about caring for persons with Down syndrome. | 83% | 17% | |
| The e-consultation confirmed that I am doing the right things in caring for persons with Down syndrome. | 67% | 11% | 22% |
| The e-consultation reassured me in my ability to care for persons with Down syndrome. | 94% | | 6% |
| The e-consultation reminded me of something I already knew about caring for persons with Down syndrome. | 100% | | |
| I was dissatisfied with the e-consultation. | | 100% | |
| There was a problem with the information in the e-consultation. | | 100% | |
| I disagreed with the content of the e-consultation. | 6% | 94% | |
| I think the e-consultation was potentially harmful. | | 100% | |
| The e-consultation had no impact at all on me or my practice. | | 100% | |

Example: ELECTRONIC CONSULTATION

Syndrome Specific Screening Tests and Evaluations:

Tests: TSH, Echocardiography. (Celiac antibody screening conducted in 2013)

Referrals: Audiology, Dentistry. (Ophthalmology exam in 2015)

Physical Examination: Examine for and remove cerumen impactions. Examine oral cavity for dental disease, gingivitis, periodontitis. Screen for features of cervical myelopathy by assessing for lower extremity hyper-reflexia and ankle clonus.

Pharmacotherapy Review:

Levothyroxine: Annual TSH was ordered, but patient did not show for blood draw.

Vitamin D supplementation: Universal vitamin D supplementation is recommended for persons with Down syndrome, typically 2000 IU of vitamin D3, in the absence of kidney disease.

Immunizations:

PCV-13 followed by Pneumovax-23 are advised because of observed increased risk for pneumococcal bacteremia associated with Down syndrome.

Community and On-line Resources:

Support, Advocacy and Education: The UpSide of Downs: 216-447-1955

Future Planning: <https://futureplanning.thearc.org>: Free web-based system to help individuals with developmental disabilities and their families plan and prepare for the future.

Helpful books for families and caregivers include: ***Mental Wellness in Adults with Down Syndrome*** by: Dennis McGuire Ph.D & Brian Chicoine MD and ***Guide to Good Health for Teens & Adults with Down Syndrome*** by: Brian Chicoine MD & Dennis McGuire Ph.D.

Recreation and Socialization: Mayfield Village, where **** resides, has community-based supported activities for persons with developmental disabilities. Additional resources for recreation and socialization can be found on **The Arc of Greater Cleveland** website <http://thearcofgreatercleveland.org/>.

Best Buddies is a national organization with local chapters which creates opportunities for one-to-one friendships, integrated employment and leadership development for people with intellectual and developmental disabilities (IDD). Local chapter is through Case Western Reserve University, contact emilyreed@bestbuddies.org

Transition-Age Considerations:

Transition-related issues include: Application for Supplemental Security Income (SSI); application for Medicaid; determination of job-readiness vs. additional formal education/training; review of Individualized Education Plan (IEP) to assure there are adequate accommodations, if further formal education/training is planned.

Clinical Rationale for Virtual Consultation Recommendations:

Echocardiography is recommended ONCE in adults with Down syndrome to assess for valvular disease, which can emerge in adulthood in persons with Down syndrome.

Hearing impairment is common in Down syndrome and audiometry is recommended every 2 years.

Oral disease, including dental caries, gingivitis, and periodontitis is more common in Down syndrome. Examination by dentist is recommended every 6 months.

Osteoporosis is more common in Down syndrome. Individuals should have optimum vitamin D intake, typically requiring supplementation of vitamin D3 2000 IU daily.

Testicular cancer occurs 5 X more frequently in men with Down syndrome compared to general population.

Example: ELECTRONIC CONSULTATION

Syndrome Specific Screening Tests and Evaluations:

Tests: Tissue transglutaminase IgA, serum IgA. [TSH, Echocardiography are up-to-date.]

Referrals: Ophthalmology, Dentistry, Audiology

Physical Examination: Examine for and remove cerumen impactions. Examine oral cavity for dental disease, gingivitis, periodontitis. Screen for features of cervical myelopathy by assessing for lower extremity hyper-reflexia and ankle clonus. Examine scrotum for testicular masses.

Screening for Dementia: Because of risk for early-onset dementia, individuals with Down syndrome should undergo screening for dementia beginning age 40 by inquiring about changes in adaptive functioning, cognition, and behavior. An Early Detection and Screen for Dementia instrument can be accessed at <http://aadmd.org/ntg/screening>.

Chronic Disease Management:

Class III Obesity (BMI/ 42 kg/m2):

Individuals with Down syndrome can achieve a healthy body weight. *****'s morbid obesity is a serious health issue which will markedly shorten his life expectancy, if untreated.

He should be referred to a registered dietician. Weigh weekly. Set weight loss goals. Include specific weight loss targets as part of the health goals for his Individual Service Plan (ISP).

***** and his family should be referred to The Arc of Greater Cleveland (216-622-0755) for enrollment in their **Healthy Lifestyles program**. A new program cycle is beginning now, in the winter of 2017.

Other resources to assist ***** in adopting a healthy lifestyle include: SOAR (Sports Opportunities and Active Recreation), contact Scott Montgomery 440-327-6454; Special Olympics.

Mental Health: Emotional distress in individuals with Down syndrome is expressed in ways that are determined by their developmental stage and life experiences. Self-talk and "psychoform" verbalizations may be mistaken clinically as psychotic features.

Anxiety Disorders and anxiety-associated behaviors are common in persons with Down syndrome. These include features of generalized anxiety disorder, panic disorders, phobias, agoraphobias, and social anxiety.

Delayed and prolonged and atypical **grief responses** are often observed in persons with Down syndrome.

Post-traumatic stress phenomena may manifest as peculiar and bizarre verbalizations and behaviors. Individuals with Down syndrome may recount distressing life events in a perseverative fashion; events that occurred long ago may be recalled as if they were recent.

History of Gout:

Individuals with Down syndrome are prone to gout because of poor renal excretion of uric acid. There is no measurement of serum uric acid in EMR. DEMADDEX could worsen hyperuricemia and contribute to gouty attacks.

Diagnostic Evaluations:

Risk for Obstructive Sleep Apnea: ***** should undergo evaluation for OSA if records of previous sleep studies cannot be secured. Approximately 50% of adults with Down syndrome have some degree of OSA. *****'s risk is much greater, given his morbid obesity. A home-based screening test might be the easiest way to begin evaluation.

Immunizations:

PCV-13 is advised because of observed increased risk for pneumococcal bacteremia associated with Down syndrome.

Hepatitis B vaccination series should be completed (or confirmed historically) because of increased risk for chronic HBV infection in persons with Down syndrome if infected with hepatitis B.

Community and On-line Resources:

Support, Advocacy and Education: The UpSide of Downs: 216-447-8763.

Future Planning: <https://futureplanning.thearc.org>: Free web-based system to help individuals with developmental disabilities and their families plan and prepare for the future.

Helpful books for families and caregivers include: **Mental Wellness in Adults with Down Syndrome** by: Dennis McGuire Ph.D & Brian Chicoine MD and **Guide to Good Health for Teens & Adults with Down Syndrome** by: Brian Chicoine MD & Dennis McGuire Ph.D

Recreation and Socialization resources can be found on The Arc of Greater Cleveland website <http://thearcofgreatercleveland.org/>.

Best Buddies is a national organization with local chapters which creates opportunities for one-to-one friendships, integrated employment and leadership development for people with intellectual and developmental disabilities (IDD). Local chapter is through Case Western Reserve University, contact emilyreed@bestbuddies.org

Quantum Leap is a east-side resource for recreation: 216-696-2716.

Jewish Community Center offers access of aquatic exercise.

The Arc of Greater Cleveland can provide assistance to his family regarding resources for activity, nutrition and support systems.

Clinical Rationale for Virtual Consultation Recommendations:

Celiac disease is estimated to be as much as 10 X more prevalent in persons with Down syndrome compared to the general population. Celiac disease may be mistaken as IBS, both in the general population and in persons with Down syndrome. Many authorities suggest periodic screening for celiac disease by antibody testing (e.g., tissue transglutaminase IgA Ab) every 3-5 years. Symptom-triggered celiac disease screening misses many individuals with Down syndrome who develop celiac disease.

Dysmotility Disorders are very common in Down syndrome and may manifest anywhere throughout the GI tract, especially in individuals who have had prior GI surgeries.

Echocardiography is recommended ONCE in adults with Down syndrome to assess for valvular disease, which can emerge in adulthood in persons with Down syndrome.

Gallbladder Disease: The risk of gallbladder disease may be 3 X that of the general population.

Gout is more common in persons with Down syndrome due to impaired renal excretion of uric acid.

Hearing impairment is common in Down syndrome and audiometry is recommended every 2 years.

Hip disease associated with Down syndrome includes unilateral or bilateral subluxation, degenerative arthritis, dysplasia, and slipped capital femoral epiphysis. In one case series, 28% of adults with Down syndrome had significant hip pathology.

H pylori infections are very common in persons with developmental disabilities, including Down syndrome.

IgA levels are measured ONCE to assure no selective IgA deficiency, which is more common in persons with Down syndrome. Selective IgA deficiency would render IgA-based celiac antibody testing falsely negative.

Intestinal Parasitosis is more common in persons with Down syndrome, attributed to both poor hygiene and to impaired immune system function.

Oral disease, including dental caries, gingivitis, and periodontitis is more common in Down syndrome. Examination by dentist is recommended every 6 months.

Osteoporosis is more common in Down syndrome. Additional risk factors include chronic use of PPIs. Individuals should have optimum vitamin D intake, typically requiring supplementation of vitamin D3 2000 IU daily. Given his chronic kidney disease, this dose may need to be adjusted, according to blood levels of 25-hydroxy-vitamin D.

Spinal Disease: In addition to atlanto-axial instability, because of the generalized ligamentous laxity associated with Down syndrome, instability can occur in other areas of the cervical spine. Early and severe degenerative disease of the cervical spine is common. Congenital narrowing of central spinal canal predisposes to spinal stenosis.

Testicular cancer occurs 5 X more frequently in men with Down syndrome compared to general population.

Thyroid Disorders eventually occur in up to 30% of individuals with Down syndrome. Screening by TSH is recommended annually.

Visual impairment and ocular diseases are common in Down syndrome and ophthalmologic evaluation is recommended every 2 years.

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