



MASSACHUSETTS
GENERAL HOSPITAL



HARVARD
MEDICAL SCHOOL



**An online personal care plan
for caregivers and primary care physicians**

Kavita Krell, Director of Customer Experience
Kelsey Haugen, Director of Sales

We would like to tell 5 stories.

1. A story of *why* we got there
2. A story of *how* we got there
3. *What* we are building
4. *How* well are we doing
5. *What* is next?

Why we got there.

Parents who have children with Down syndrome worry the medical established has passed them by.

*"In an era when the offer of prenatal testing is universal and termination of pregnancies involving Down syndrome is commonplace, families of people with Down syndrome often say **they worry that the medical establishment has passed them by.** They feel marginalized, unseen, and unserved. Families of people with Down syndrome don't want admiration, and they don't want pity. **They feel a critical and urgent need for adequate health care for their loved ones, which is the right of every American.**"*



Patricia E. Bauer
Mother

—Patricia Bauer, caregiver

71 Down syndrome clinics in U.S.



www.ndss.org/Resources/Health-Care

At best, though, all of these clinics serve
< 5% of the population with Down syndrome.

My sister is one of those patients who does not have access to a specialty clinic.

- I have a sister with Down syndrome.
- She is 25 years old.
- There are no adult Down syndrome clinics where she lives



It is unreasonable & impractical for primary care physicians to stay up-to-date on Down syndrome medical care.

<10% of patients with Down syndrome were up-to-date on 5 of the basic healthcare screens recommended by the American Academy of Pediatrics, according to our study

RESEARCH ARTICLE

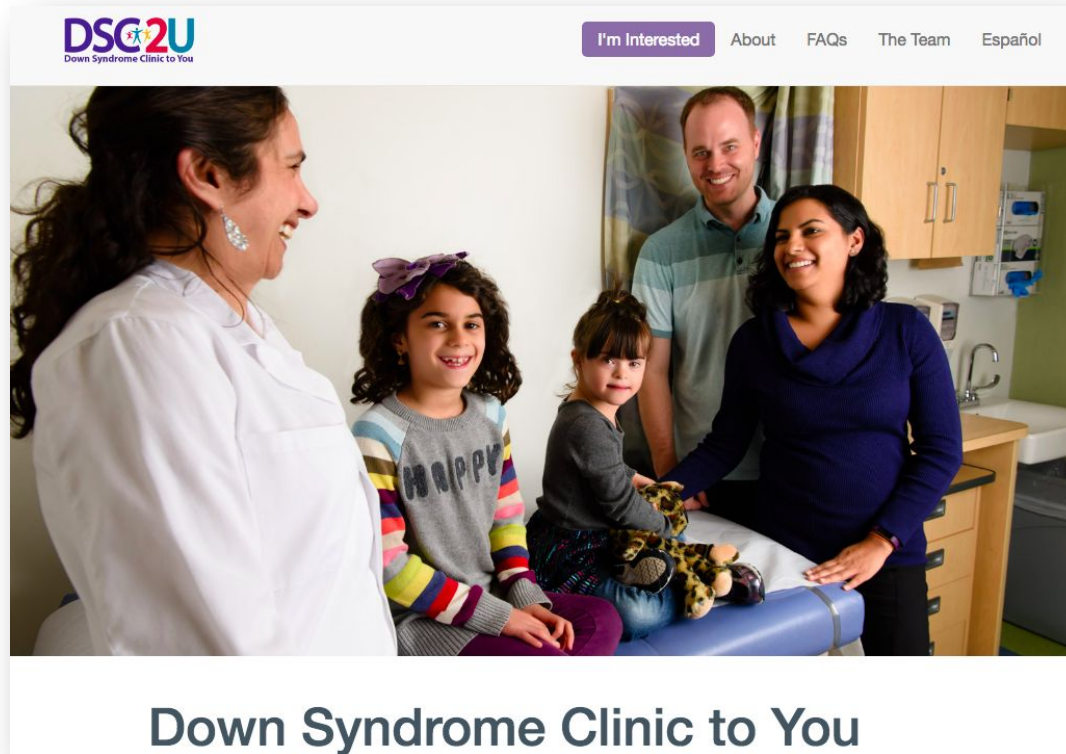
AMERICAN JOURNAL OF
medical genetics PART
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Contributions of a Specialty Clinic for Children and Adolescents with Down Syndrome

Brian G. Skotko,^{1*} Emily Jean Davidson,² and Gil S. Weintraub³

How we got there.

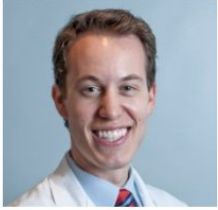
We just completed a 3-year, \$2.1M project, funded by PCORI.



MassGeneral Hospital DSC2U Team



Physicians



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MD, MPP
Director



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Medical Informatics
Lab of Computer Science



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Dyan Blewett



Stephen Lorenz



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PhD



Mark Wylie



Grace Hsieh
RN, PhD



Holly Parker



Lloyd Clarke

National Advisory Groups Provided Critical Inputs

Expert Advisory Workgroup



Marilyn Bull, MD



Brian Chicoine, MD



Karen R. Sepucha, PhD



Melissa A. Parisi, MD, PhD



Allie Schwartz, MD

PCP Advisory Workgroup



Ariel S. Frey-Vogel, MD



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Caregiver Advisory Workgroup



Sandra Baker
Mother



Patricia E. Bauer
Mother



Sarah Cullen
Mother



Melissa Kline Skavlem
Mother



Lisa Majewski, MSW
Mother



Jawanda Barnett Mast
Mother



Maureen Gallagher
Mother

What we are building

**DSC2U is an automated, direct-to-consumer tool
in English & Spanish**



dsc2u.org

1 General Information

2 Current Medical Concerns

3 Current Mental Health/Behavioral Concerns

4 Immunizations

5 Nutrition

6 Past Medical History

7 Blood work and Diagnostic Testing

8 School and Therapies

9 Life Skills Checklist

10 Down Syndrome Community

Virtual Clinic (October 5th version)

General Information**Information About the Patient****First name** *(required)*

Kristin

Date of birth *(required)*

08/20/1980

Gender *(required)*

- Male
- Female
- Other

What is Kristin's diagnosis? Please click any and all that may apply.

- Trisomy 21
- Mosaic Down Syndrome

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Virtual Clinic (October 5th version)

Current Medical Concerns

Are any of these symptoms a current concern for Kristin? Please select any and all that may Kristin may have experienced within the last month.

General

Fever that hasn't gone away in at least five days

Weight gain

Undesired weight loss

New limp

Increasing fatigue

Eyes

Concerns about vision

Involuntary eye movements

DSC2U available in Spanish, too

1 Información general

2 Afecciones médicas actuales

3 Inquietudes actuales sobre la salud mental o la conducta

4 Análisis de sangre y pruebas de diagnóstico

5 Antecedentes médicos

6 Inmunizaciones

7 Nutrición y deglución

8 Escuela, terapias y recursos

9 Lista de referencia sobre habilidades para la vida

10 Comunidad del síndrome de Down

11 Comentarios

Down Syndrome Clinic to You (DSC2U)

Inquietudes actuales sobre la salud mental o la conducta

¿Alguno de estos síntomas le provocó una inquietud a Molly durante los últimos 6 meses?

Seleccione todas las opciones que Molly pueda haber experimentado durante los **ÚLTIMOS SEIS MESES**.

- Sensación de desánimo, depresión o desesperanza
- Poco interés en realizar actividades o poco placer al realizarlas
- Lloro fácilmente sin ningún motivo
- Se desplaza lentamente
- Pérdida de las habilidades adquiridas previamente
- Come demasiado o come muy poco
- Problemas para conciliar el sueño o mantenerse dormido
- Está tenso, ansioso, preocupado
- Le angustia estar solo

Saving...

Down Syndrome Clinic to You (DSC2U)

Nutrition and Swallowing

1 General Information

2 Current Medical Concerns

3 Current Mental Health/Behavioral Concerns

4 Bloodwork and Diagnostic Testing

5 Past Medical History

6 Immunizations

7 Nutrition and Swallowing

8 School, Therapies, and Resources

9 Life Skills Checklist

10 Down Syndrome Community

11 Feedback

Types of food eaten

How many cups of sugar-sweetened drinks does Kristin have in an average day?

- Less than 1 per day
- More than 1 per day
- Not sure

This includes: juice, sports drinks, soda, chocolate milk, etc. This does not include artificially sweetened drinks like Crystal light and diet sodas.

How many servings of calcium-rich food does Kristin have in a typical day?

- 2 or less per day
- 2 or less per day, plus a calcium chew or multi-vitamin with calcium
- 3 or more per day
- Not sure

1 serving = 1 cup of milk, or fortified soy or almond milk, 1 ounce of cheese (size of 3 die), 1/2 cup of yogurt, 1 cup of leafy greens. Some examples of calcium rich foods include: dairy milk, fortified soy or almond milk, tofu, cheese, yogurt, leafy green vegetables.

Do any of the following describe Kristin's snack preferences?

- Eats mostly "munchie" food like chips and pretzels
- Eats mostly sweet foods like candy, baked goods, and ice cream

1 General Information

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6 Past Medical History

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Virtual Clinic (October 5th version)

Blood work and Diagnostic Testing

Has Kristin...

...seen an audiologist for a formal hearing examination in the past 2 years?

- Yes
- No
- Not sure

...seen an ophthalmologist for a formal eye examination in the past 2 years?

- Yes
- No
- Not sure

...ever had a sleep study performed for obstructive sleep apnea?

- Yes
- No
- Not sure

1. Caregiver Checklist for You

2. Primary Care Provider Plan to Share

Personalized Checklist for Molly's Caregiver

Thank you very much for taking the time to complete the Down Syndrome Clinic to You (DSC2U) intake form. **We used your responses to create this personalized checklist of information, resources, and recommendations for Molly.** To find out why certain suggestions were made, please click on the "**Why?**" link after each recommendation.

We know Molly's health and wellness are very important to you, and we hope these resources will be helpful as you oversee her care. At the same time, a long list can sometimes feel a bit overwhelming. We encourage you to talk through the contents of this checklist with Molly's primary care provider and other trusted caregivers or family members who may help you with Molly's care.

The suggestions in this checklist were generated by a computer, based on your answers to the DSC2U intake form. The checklist was not prepared or reviewed by a clinician specifically for Molly but was assembled from information that was carefully chosen by a team of medical experts. Whenever possible, the information in this document was drawn from national healthcare guidelines for people with Down syndrome. The information has been reviewed to make sure it is consistent with current best practices.

Endocrine Conditions & Down Syndrome

Individuals with Down syndrome have a higher incidence of endocrine problems than the general population. The endocrine system refers to a set of glands that include the thyroid, adrenal and pituitary glands.

What Is Hypothyroidism?

Hypothyroidism results from a malfunctioning thyroid gland. The thyroid gland is involved in various metabolic processes controlling how quickly the body uses energy, makes proteins and regulates hormones. In hypothyroidism, the synthesis of the hormone thyroxin is decreased. Thyroxin is the hormone that promotes growth of the brain and other body tissue.

How Common Is Hypothyroidism?

Hypothyroidism is the most common endocrine problem in children with Down syndrome. It is estimated that approximately 10% of children with Down syndrome have congenital or acquired thyroid disease. Hypothyroidism is also common in adults with Down syndrome and can lead to symptoms of fatigue, mental sluggishness, weight fluctuations and irritability. Studies of adults with Down syndrome vary widely, but the incidence of thyroid disease in adults with Down syndrome is believed to be between 13% and 50%. Hypothyroidism can occur at any time from infancy through adulthood.

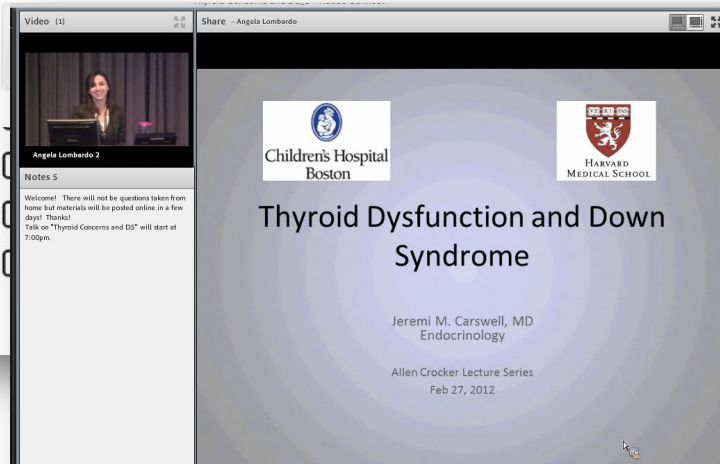
Labs, Tests, and Procedures for Molly

It is recommended that you ask Molly's doctor to consider ordering the following during the up

- Eye exam.** Molly is due for a vision test by an eye doctor specialist. [More info on eye exams \[video\]](#). [Why?](#)
- Celiac screen** (blood work). Molly has symptoms that might be consistent with celiac disease. [Follow-up testing \[video\]](#). Treatment options are available for those with a confirmed diagnosis. [Why?](#)
- Sleep study.** Molly has symptoms that might be consistent with obstructive sleep apnea. [More info on sleep studies \[video\]](#). Here is how caregivers can prepare for sleep studies. [Here is a social story that Molly can use to prepare for the sleep study.](#) [Why?](#)
- Thyroid function tests** (blood work). Molly is due for her thyroid check. Further testing is needed. [Treatment options are available for those with a confirmed diagnosis \[video\]](#). [More information on thyroid conditions in Down syndrome.](#) [Why?](#)

is drawn. Individuals with Down syndrome have a significant risk of developing thyroid problems, which are important to check annually.

having Molly referred to a Speech and Language Pathologist and a neuropsychologist to



Video (1) Share - Angela Lombardo

Angela Lombardo 2

Notes 5

Welcome! There will not be questions taken from home but materials will be posted online in a few days. Thank! Talk on "Thyroid Concerns and DS" will start at 7:00pm.

Children's Hospital Boston HARVARD MEDICAL SCHOOL

Thyroid Dysfunction and Down Syndrome

Jeremi M. Carswell, MD
Endocrinology

Allen Crocker Lecture Series
Feb 27, 2012

Woodbine House

Publisher of the Special-Needs Collection...books for parents, children, teachers, and other professionals

Home Shop All Down Syndrome Autism ADD & ADHD Children's Books Sale

Home > Down Syndrome > Mental Wellness in Adults with Down Syndrome: A Guide to Emotional and Behavioral Strengths and Challenges

Mental Wellness in Adults with Down Syndrome



Dennis McGuire, Ph.D. & Brian Chicoine, M.D.

Mental Wellness in Adults with Down Syndrome: A Guide to Emotional and Behavioral Strengths and Challenges

\$24.95

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Nutrition Recommendations for Molly

People with Down syndrome, in general, have slower metabolism. Focusing on healthy eating, at all ages, is important. Sometimes, making just a few tweaks in one's diet can go a long way. Based on your responses, here are some suggestions:

- Ask Molly's doctor if she should have **more calcium in the diet**. [This handout](#) details daily calcium needs based on age, and foods you can offer to ensure that Molly is meeting her needs. Also, individuals should take between 400 - 1000 international units of Vitamin D a day based on their age, which can be consumed through food, pill, or supplement. [Why?](#)
- Healthier snacking**. [This handout](#) is a great visual guide to encourage healthy snack choices. [Here are some options for healthy, low-calorie snacks.](#)
[Why?](#)
- Consider reading [this book](#) for a detailed overview on **healthy nutrition for people with Down syndrome**. [Why?](#)

Life
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Goals

DOWN SYNDROME PROGRAM
www.massgeneral.org/downsyndrome



Using Public Transportation on My Own



DOWN SYNDROME PROGRAM
www.massgeneral.org/downsyndrome



This is a good time for me to learn where to pay for or buy a ticket for my ride.

I am going to _____

My trip will cost me _____

I will pay for my trip with _____



book



life

Education/Therapy Resources for Molly

Recursos de educación/terapia para Molly

Cada año, los investigadores entienden mejor cómo aprenden las personas con síndrome de Down. Estos son algunos recursos específicos, basados en su solicitud, para Molly.

- Habilidades del habla y la comunicación.** Consulte el capítulo 6 de este libro [🔗](#). Puede buscar fonoaudiólogos locales [aquí](#) [🔗](#).
- Habilidades de adaptación para la vida.** Consulte el capítulo 9 de este libro [🔗](#).
- Habilidades vocacionales/oportunidades de empleo.** Haga clic aquí para ver un resumen de las opciones de empleo [🔗](#). Además, consulte los recursos de la campaña [Your Next Star](#) [🔗](#), que incluyen [recursos para los solicitantes](#) [🔗](#) y [empleadores](#) [🔗](#). Estos son más recursos e historias de éxito de [#DSWORKS@](#) [🔗](#). Para obtener más información sobre el empleo, consulte [este sitio web](#) [🔗](#) y [este manual](#) [🔗](#).

Información y recursos para el cuidador de Molly

Estos son algunos recursos específicos basados en su solicitud.

- DS-Connect®.** Considere inscribir a Molly en [DS-Connect](#) [🔗](#) para que esté en contacto con investigadores y proveedores de servicios de salud. [¿Por qué?](#)
- Global Down Syndrome Foundation.** Encuentre más información en su sitio web. [🔗](#) Suscríbese a su lista de correo. [🔗](#) [¿Por qué?](#)
- LuMind Research Down Syndrome Foundation.** Encuentre más información en su sitio web. [🔗](#) Suscríbese a su lista de correo. [🔗](#) [¿Por qué?](#)
- National Down Syndrome Congress (NDSC).** Encuentre más información en su sitio web. [🔗](#) Suscríbese a su lista de correo. [🔗](#) [¿Por qué?](#)
- National Down Syndrome Society (NDSS).** Encuentre más información en su sitio web. [🔗](#) [¿Por qué?](#)
- Organización local de síndrome de Down.** Puede acceder desde [este enlace](#) [🔗](#) para encontrar una lista con todas las organizaciones de síndrome de Down en Estados Unidos. Podrá ver si hay una cerca de usted. [¿Por qué?](#)
- Ingreso Suplementario de Seguridad Social (SSI).** Consulte [este kit para principiantes](#) [🔗](#) para los adultos con discapacidades que solicitan beneficios a la seguridad social. Además, consulte [este sitio web](#) [🔗](#) para ver una guía paso a paso sobre cómo solicitar la SSI. [¿Por qué?](#)

1. Caregiver Checklist for You

2. Primary Care Provider Plan to Share



MASSACHUSETTS
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HARVARD
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07/08/2018

Dear Dr. Schwartz,

The caregiver of your patient, **Molly Schmidt (DOB 08/20/1980)**, is participating in a research project aimed at improving healthcare outcomes for patients with Down syndrome. The project is coordinated by a team of clinicians and researchers at Massachusetts General Hospital in Boston and is funded by a grant from the Patient-Centered Outcomes Research Institute. More information about our project can be found at www.dsc2u.org.

The caregiver of your patient completed an online questionnaire which generated automated suggestions for testing and treatment that are anchored on published practice guidelines and/or expert consensus. The online tool encouraged **Molly's** caregiver to share and discuss these with you.

On the next page, you will find a checklist of these recommendations and list of educational resources specific for Down syndrome. We hope you will find them helpful during your next clinical visit with this patient.

Sincerely,

MGH Down Syndrome Program Research Team

Recommended Labs, Tests, and Procedures for Molly

(AAP = American Academy of Pediatrics)

- Ophthalmology Evaluation:** Experts recommend an ophthalmologic exam at least every two years for adults with Down syndrome, ages 21 and older.
- Celiac screen** (total IgA & TTG-IgA). Molly's caregiver indicated that within the past month of completing our intake, she had constipation that is hard to treat, nausea, passing excessive gas, bloating, mood swings, crying easily or for no reason. Molly has never had celiac testing done. Celiac disease is more common in people with Down syndrome.
- Sleep study.** Molly's caregiver indicated that within the past month of completing our intake, Molly experienced snoring, gasping, snorting, choking during sleep, sleeping on multiple pillows, not feeling refreshed despite adequate sleep, feeling down, depressed, or hopeless, crying easily or for no reason, moving slowly, distress about being alone. Molly has also never had a sleep study. Experts recommend that everyone with Down syndrome be evaluated for sleep apnea if symptoms arise. We would recommend a sleep study to assess for obstructive sleep apnea, which can occur in up to 75% of people with Down syndrome. Research has also shown that patients with Down syndrome may lose up to 9 IQ points within a year when their apnea is untreated.
- Thyroid function tests** (TSH & fT4). Molly's caregiver indicated that within the past 12 months of completing our intake, Molly has not had her thyroid levels drawn. Experts recommend that everyone with Down syndrome have their thyroid levels checked once annually, given the high incidence of thyroid disorders in patients with Down syndrome.

Potential New Conditions/Diagnoses to be Considered for Molly

We recommend that you consider initiating evaluations for the following conditions that can often co-occur with Down syndrome:

- Depression.** Molly's caregiver marked that within the past 6 months of completing our intake, Molly has experienced feeling down, depressed, or hopeless, little interest or pleasure in doing things, crying easily for no reason, moving slowly, distress about being alone. Generalized depression is a common co-occurring condition for people with Down syndrome. Resources and book recommendations are on our web page. Psychopharmacological consult should be considered if possible underlying medical causes can be eliminated.
- Arthropathies.** Molly's caregiver indicated that within the past month of completing our intake, Molly has experienced joint stiffness, joint pain. Gout and osteoarthritis are all common in people with Down syndrome, and a full standard work up should be pursued in patients presenting with symptoms of joint pathology, including a workup for other connective tissue disorders.
- Chronic constipation.** Molly's caregiver indicated that within the past month of completing our intake, Molly experienced constipation that is hard to treat. Chronic constipation is a common feature in patients with Down syndrome and can lead to behavioral issues if left untreated. Consider obtaining a KUB and trying medication to alleviate the constipation if needed.

OTHER INFORMATION, RESOURCES, and SUPPORTS for Molly

For your information, we provided Molly's caregiver links to the following online resources and information. If you would like online access to these resources, please go to <https://www.dsc2u.org/primarycare>.

Health and Wellness Resources

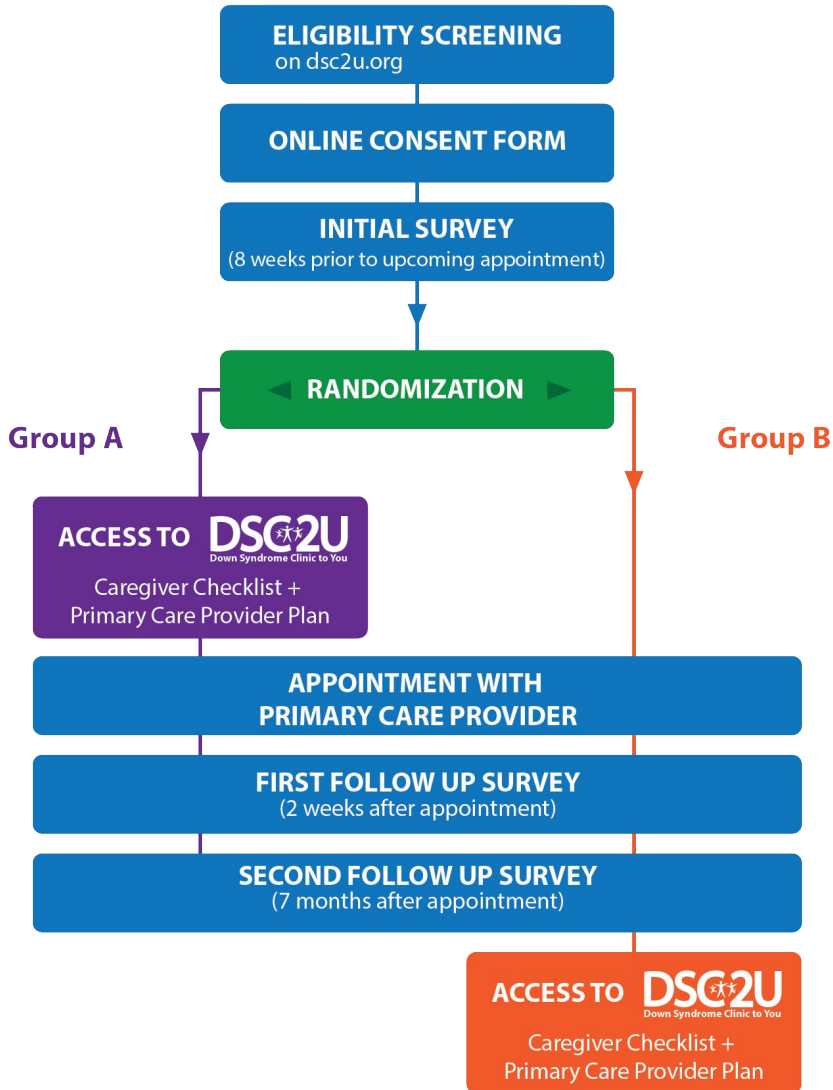
- Disruptive behaviors
 - Oral-Contraceptive Pills (OCPs) for women with Down syndrome
 - Dating and Social Boundaries
 - Puberty and Sexual Development
 - Resources for weight gain in people with Down syndrome
-

Nutrition

- Molly should be encouraged to increase her overall calcium intake.
 - Molly should be encouraged to develop healthier snacking habits.
 - Resources for healthy eating habits in persons with Down syndrome.
-

How well are we doing?

We conducted a national randomized control trial to assess efficacy and satisfaction



Specific Aim 1: To test whether a customizable Caregiver *Plan* and PCP *Letter* versus usual care will increase caregiver-reported, provider-driven health actions consistent with national guidelines.

Specific Aim 2: To determine whether a customizable *Plan* and *Letter* versus usual care is satisfactory to caregivers and providers and improves quality of life for caregivers/families.

DSC2U is effective

The group that received DSC2U had a **1.6-fold increase** in the number of indicated evaluations that were recommended by the primary care provider or completed compared to controls.

© American College of Medical Genetics and Genomics

ARTICLE | Genetics
inMedicine



A randomized controlled trial of an online health tool about Down syndrome

Jeanhee Chung, MD^{1,2}, Karen Donelan, ScD, EdM^{2,3}, Eric A. Macklin, PhD⁴, Alison Schwartz, MD^{5,6}, Ibrahim Elsharkawi, MD⁵, Amy Torres, BS⁵, Yichuan Grace Hsieh, PhD, RN^{1,2}, Holly Parker, BA¹, Stephen Lorenz, BA¹, Vasiliki Patsiogiannis, BA⁵, Stephanie L. Santoro, MD^{5,6}, Mark Wylie, BA¹, Lloyd Clarke, BA¹, Greg Estey, BA¹, Sandra Baker⁷, Patricia E. Bauer, BGS⁸, Marilyn Bull, MD⁹, Brian Chicoine, MD¹⁰, Sarah Cullen, BA¹¹, Ariel Frey-Vogel, MD, MAT⁶, Maureen Gallagher, MS¹¹, Reem Hasan, MD, PhD¹², Ashley Lamb, MD, MPH¹³, Lisa Majewski, MSW, Jawanda Mast, MS, Travis Riddell, MD, MPH¹⁴, Karen Sepucha, PhD¹⁵, Melissa Skavlem, BA and Brian G. Skotko, MD, MPP^{5,6}

<https://rdcu.be/b6Hk4>

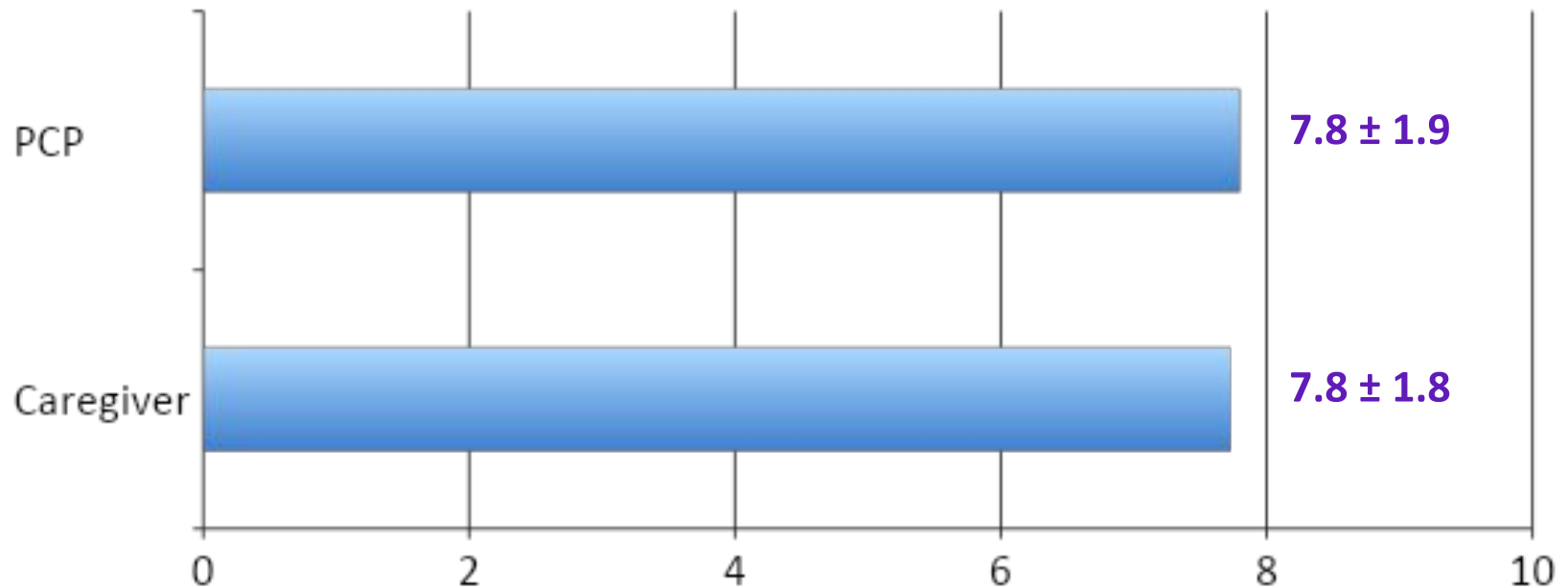
Caregivers are highly satisfied with DSC2U

Question	Yes, Definitely	Yes, Somewhat	No
Did Caregiver Checklist explain recommendations in a way that was easy for you to understand?	86%	11%	3%
Would you recommend the DSC2U to another caregiver of someone with Down syndrome?	83%	17%	0%
Did you use the links to information that were included in the Caregiver Checklist?	38%	39%	24%

PCPs are satisfied with DSC2U

Question	Yes, Definitely	Yes, Somewhat	No
Did you discuss this primary care plan or any of its recommendations with the caregivers?	75%	25%	0%
Did you agree with the recommendations in this Plan?	61%	36%	3%
Were you interested in any of the information in this Plan?	71%	29%	0%

Overall, DSC2U had high rates of satisfaction from caregivers & PCPs



Using any number from 0 to 10, where 0 is the LEAST HELPFUL information possible and 10 is the MOST HELPFUL information possible, what number would you use to rate the CAREGIVER CHECKLIST or PRIMARY CARE PROVIDER PLAN?

What is next?

DSC2U has been launched!



Founding Philanthropic Supporter

dsc2u.org

All families across the globe can access DSC2U right now in English and Spanish

DSC2U
Down Syndrome Clinic to You

Login Español

Caregivers Providers Organizations Insurers Contact

MASSACHUSETTS GENERAL HOSPITAL
MassGeneral Hospital for Children
HARVARD MEDICAL SCHOOL TEACHING HOSPITAL

DSC2U

A personalized care plan

We're here to help you. We know it may not be easy to get expert medical care for your loved one with Down syndrome.

dsc2u.org \$49 for each usage

DSC2U can serve as a blueprint for many other diseases/conditions

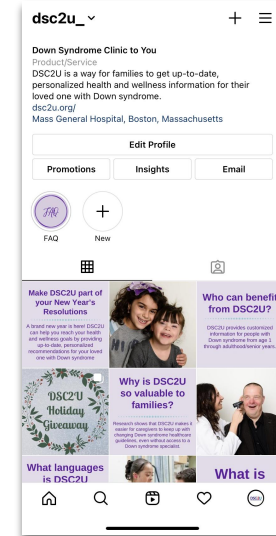
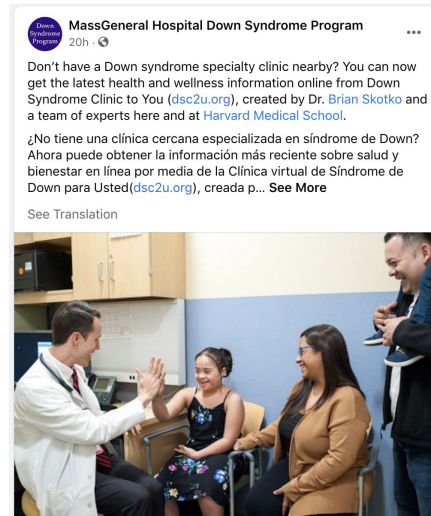
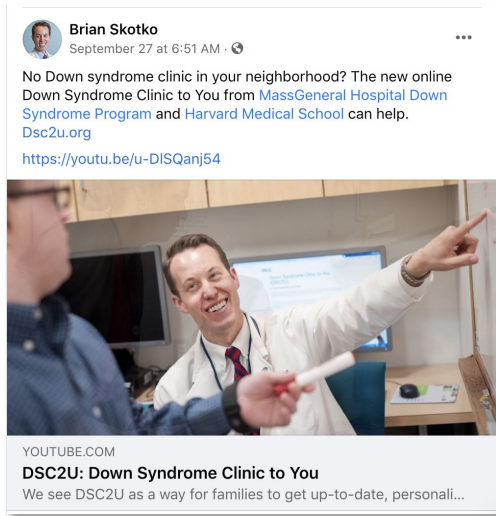
We have already been contacted by physician experts in

- Marfan syndrome
- Adult Congenital Heart Disease
- Pediatric Allergies

In short, any disease/condition with a medical playbook that needs management within primary care settings can benefit from our product.

This would allow high-cost specialty care to be delivered in cost-effective settings (PCP offices).

Please help us spread the word about DSC2U



[facebook.com/drbrianskotko](https://www.facebook.com/drbrianskotko) www.facebook.com/MGHDownSyndrome/ www.instagram.com/dsc2u_/



twitter.com/MGHDownSyndrome

twitter.com/brianskotko

Thank you

KAVITA KRELL and KELSEY HAUGEN

E-mail: dsc2u@mgh.harvard.edu

Web page: www.dsc2u.org

Facebook: <https://www.facebook.com/MGHDownSyndrome/>

Twitter: <https://twitter.com/mghdownsyndrome>

Instagram: [@dsc2u_](https://www.instagram.com/dsc2u_)

Clinic: www.massgeneral.org/downsyndrome

Research: www.massgeneral.org/downsyndromeresearch