



**GLOBAL**  
DOWN SYNDROME FOUNDATION®

# **Global Medical Care Guidelines for Adults with Down Syndrome: A Legacy Supporter Update!**

**Michelle Sie Whitten, President & CEO**

**Bryn Gelaro, LSW, Director of Adult Initiatives & Special Projects,  
Co-Author on guidelines**

*December 12, 2019*

# A Quick Overview

- ❖ **Happy Holidays and Welcome!**
- ❖ **Global Down Syndrome Foundation**
  - An introduction to Global's work
  - RESEARCH, MEDICAL CARE, EDUCATION & ADVOCACY
  - The importance of Research & Medical Care
- ❖ **Medical Care Guidelines for Adults with DS Update:**
  - Success stories (already)
  - PICOTS & Grade System framework
  - Results from our focus groups
  - Managing Expectations - what the guidelines do and do not include
  - Overachieving! Possible Toolkits...
- ❖ **NEXT STEPS for the Guidelines**
  - THANK YOU to our Supporters & Members!



# The Inspiration behind Global



*Anna and John J. Sie*



*Sophia & Patrick Whitten*

“Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has.”

Margaret Mead



# Global Down Syndrome Foundation

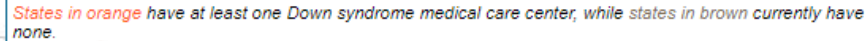
## A Unique Affiliate Model

The Global Down Syndrome Foundation is part of a network of affiliate organizations that work closely together on a daily basis to deliver on our mission - Significantly improve the lives of people with Down syndrome through ***Research, Medical Care, Education & Advocacy***:




- ❖ **Global**: fundraising, outreach, advocacy for Global and four Global Affiliates.
- ❖ **Sie Center**: over 1,700 unique patients from 28 states and 10 countries with 8 clinics (2 first in-kind)
- ❖ **Crnic Institute**: over 200 scientists working to elongate life and improve health outcomes for people with Down syndrome.
- ❖ **RMADC**: Leukine and other important breakthroughs.
- ❖ **Adult Clinic**: Goal is to have a world-class feeder for the Sie Center.

*Dedicated to collecting detailed info on DS clinics across the U.S.*



- Over 40 Pediatric centers
- 12 offer pediatric and adult care
- 10 serve only adults



# **RESEARCH & MEDICAL CARE:** **Why they are so important and how they are related?**

# The CRITICAL & TIMELY Need

## *A Population Explosion & A Eugenics Framework*

- ❖ **There is a population explosion of people with Down syndrome in the U.S. that requires dramatically *more* funding not less**
  - **Population** - is somewhere between 250,000 to 430,000
  - **Live Births** - have increased to 1 in 691 today from 1 in 1,000 in 2002
  - **Lifespan** - has more than doubled to 60 years from 28 years in the 1980s
  - **A Mini Population Explosion** - will happen over the next several decades due to increased live births and lifespan
  - **Societal Trends** - include a small but growing number of people with Down syndrome participating in college programs, choosing to get married, and living independently or semi-independently
- ❖ **There is a “eugenics framework” in countries like Iceland and Denmark...**

# Why are research and medical care so important?

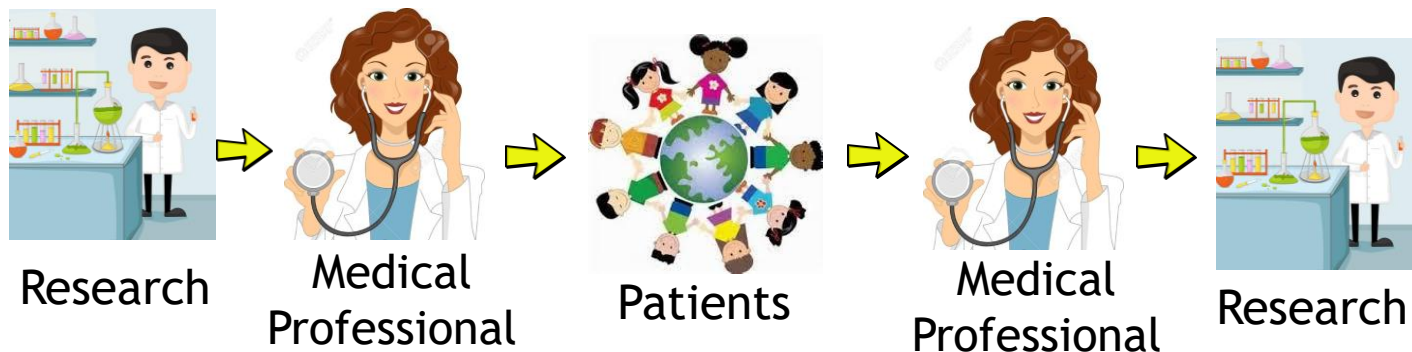
## ❖ Extends the lifespan

- In the 1980s lifespan was 28 years...today it is approximately 60
- Reasons - De-institutionalization + advances in pediatric heart surgeries

## ❖ Improves the quality of life throughout life

- 2001 “Health Supervision for Children with Down Syndrome”; current ones are a “must” and provide a great check-list (Drs. Marilyn J. Bull, William I. Cohen, Nancy Rozien)
- **No *current* guidelines for adults with Down syndrome (Drs. William I. Cohen and David S. Smith)**

## ❖ People with Down Syndrome have a Different Disease Spectrum!





# MEDICAL CARE GUIDELINES FOR ADULTS WITH DOWN SYNDROME

*An Important Legacy for our Community*



# Medical Care Guidelines for Adults with DS

## Do they already exist?

### Health Care Guidelines for Individuals with Down Syndrome

Reprinted from *Down Syndrome Quarterly*,  
Volume 4, Number 3, September, 1999

Edited by William I. Cohen M.D. for the Down Syndrome Medical Interest Group<sup>1</sup>

Dedicated to the memories of Chris Pueschel and Thomas E. Elkins M.D., two individuals, who, each in his own way, has inspired us to provide compassionate care for individuals with Down Syndrome.

#### Introduction

Individuals with Down syndrome (DS) need the usual health care screening procedures as recommended by the American Academy of Pediatrics. Adults with DS should have health evaluations using the standard accepted practices. Adults may develop certain medical problems that occur in much higher frequency than children and adults with DS. These recommendations should take into consideration the fact that the needs of individuals with DS are constantly modified as new information becomes available. Modern primary health care includes recommendations specific to these needs of individuals with DS.

#### PRACTICAL THERAPEUTICS

### Health Care Management of Adults with Down Syndrome

DAVID S. SMITH, M.D., Medical College of Wisconsin, Milwaukee, Wisconsin

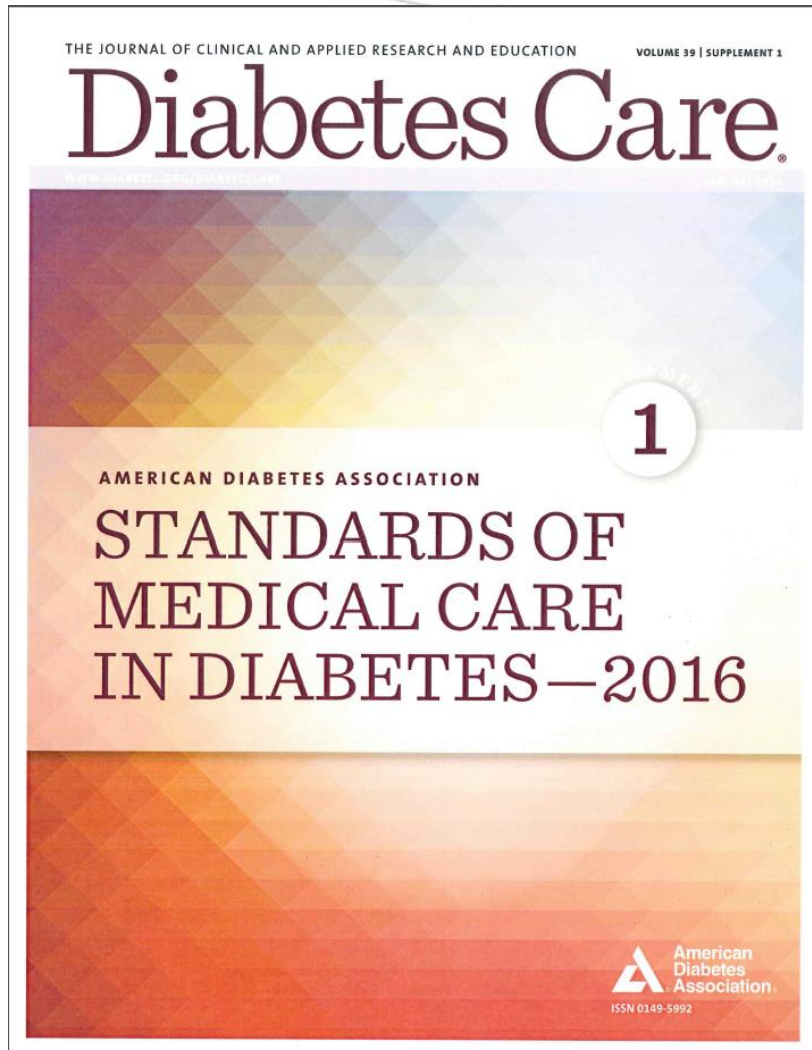
The family physician's holistic approach to patients forms the basis of good health care for adults with Down syndrome. Patients with Down syndrome are likely to have a variety of illnesses, including thyroid disease, diabetes, depression, obsessive-compulsive disorder, hearing loss, atlantoaxial subluxation and Alzheimer's disease. In addition to routine health screening, patients with Down syndrome should be screened for sleep apnea, hypothyroidism, signs and symptoms of spinal cord compression and dementia. Patients with Down syndrome may have an unusual presentation of an ordinary illness or condition, and behavior changes or a loss of function may be the only indication of medical illnesses. Plans for long-term living arrangements, estate planning and custody arrangements should be discussed with the parents or guardians. Because of improvements in health care and better education, and because more people with this condition are being raised at home, most adults with Down syndrome can expect to function well enough to live in a group home and hold a meaningful job. (Am Fam Physician 2001;64:1031-8,1039-40.)

A patient information handout on health issues in adults with Down syndrome, written by the author of this article, is on page 1039.

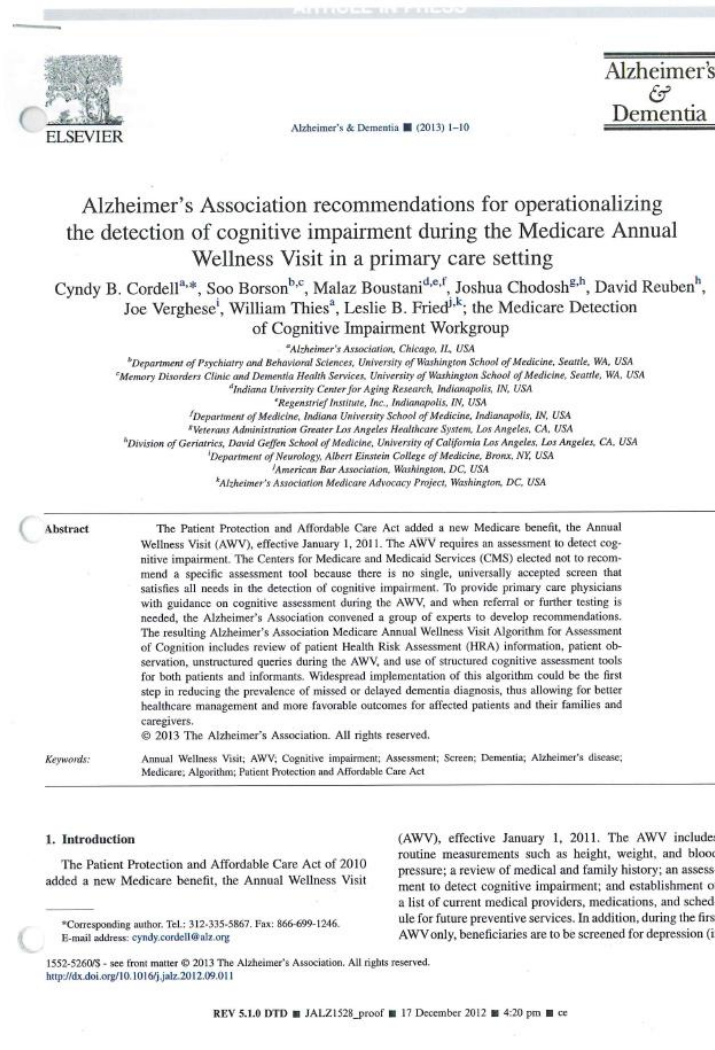
*Drs. William I Cohen and David S. Smith provided their experience, great insights, time and effort towards these publications - we should build upon their work and create a well-vetted, reviewed, periodically updated set of guidelines for today and the future...*



# Other Medical Care Guidelines



Confidential



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Adults With Down Syndrome Medical Care  
Guideline Status Update

# Medical Care Guidelines For Adults with Down Syndrome

## *Timeline*

### ❖ 2015 - 2017

- Created Global Task Force for Adults with Down syndrome; National Survey of Parents of Adults; Due diligence on national medical guidelines and national partners
- Presented our findings to the DS community at DSAIA and elsewhere; created a funding model; Recruited key adult medical experts (DSMIG is a great help); dozens of expert meetings with ECRI

### ❖ 2018

- Closed out recruitment for authors; recruited expert volunteers
- Finalized the 9 topics and conclusions on the 9 topics

### ❖ 2019-2020

- Finalize “manuscript” with ECRI and full team of authors
- Work with ECRI and authors on publication in journals and license for free through an easy on-line mechanism
- Conduct focus groups
- Start translating into the top three languages with professional medical translators
- Work with other national and local organizations for



# ECRI Institute - A National Expert

- ❖ **ECRI Institute (ECRI) is a national and international research agency and expert on creating evidence-based medical guidelines**
- ❖ **ECRI produces guidelines that are:**
  - Publishable, high quality, and renown in the medical community
- ❖ **ECRI has greater access to published literature and research than most independent medical experts**
- ❖ **They will preform the initial literature search, which is the most time-intensive component of the guideline production**
  - This will highlight research available AND the gaps and holes in the literature that need to be addressed



# Medical Care Guidelines for Adults with DS

## *The Authors*



**George Capone (MD)**- Director of Down Syndrome Clinic and Research Center, Kennedy Krieger Institute, Associate Professor, John Hopkins School of Medicine, Baltimore, Maryland. Dr. Capone attended college at Wesleyan University, worked as a research assistant at the Dana Farber Cancer Institute in Boston, obtained his medical degree from the University of Connecticut in 1983. After a residency and fellowship in pediatrics at the Children's Hospital Medical Center in Cincinnati, Dr. Capone came to Baltimore in 1988 to pursue a fellowship in neurobiology research at Johns Hopkins. Dr. Capone is also an attending physician on the institute's comprehensive rehabilitation unit.



**Brian Chicoine (MD)**- Medical Director, Advocate Medical Group Adult Down Syndrome Center, Park Ridge, Illinois. He is on the faculty of Family Medicine at Advocate Lutheran General Hospital. Dr. Chicoine is co-founder of the Adult Down Syndrome Center that has served the health and psychosocial needs of over 6, 000 adolescents and adults with Down syndrome since its inception in 1992. He graduated from Loyola University of Chicago Stritch School of Medicine and completed his Family Medicine residency at Lutheran General Hospital. He has co-authored two books "Mental Wellness of Adults with Down Syndrome," and "The Guide to Good Health for Teens and Adults with Down Syndrome" published by Woodbine House. Press.



**Barry Martin (MD)**- Assistant Professor, Division of General Internal Medicine, University of Colorado School of Medicine, Former Medical Director of the Denver Adult Down Syndrome Clinic, Denver, Colorado. He is Board Certified in Family Medicine. He has more than 20 years' experience providing primary health care for adults with disabilities, especially developmental disabilities. He served as Medical Director of the former Denver Adult Down Syndrome Clinic. Dr. Martin is a member of the National Down Syndrome Congress and the Down Syndrome Medical Interest Group.

# Medical Care Guidelines for Adults with DS

## *The Authors*



**Dennis McGuire** (PhD, LCSW)- Behavioral Expert for Adults with Down syndrome, Global Senior Consultant. He is the co-founder and former Director of Psychosocial Services for the Adult Down Syndrome Center of Lutheran General Hospital in suburban Chicago. Dr. McGuire is the co-author of *Down syndrome: Mental Wellness of Adults with Down Syndrome* (2006), and *The Guide to Good Health for Teens and Adults with Down Syndrome* (2011). He received the NDSC Theodore D. Tjossem Research Award and the World Down Syndrome Day Scientific Award. Dr. McGuire received his doctorate from the University of Illinois at Chicago and his master degree from the University of Chicago.



**Kent McKelvey** (MD)- Director of Adult and Cancer Genetics Services, University of Arkansas for Medical Sciences, Director of Winthrop P. Rockefeller Multidisciplinary Clinic for Adults with Down syndrome, Little Rock, Arkansas. He is the inaugural recipient of the Winthrop P. Rockefeller Chair in Clinical Genetics. Dr. McKelvey leads a multidisciplinary clinic that provides comprehensive services for adolescents and adults with Down syndrome. He is interested in public health issues that take into account the growing number of people with genetic syndromes and the complex interplay of medical and social needs in this population across lifespan.



**Moya Peterson**, (PhD, APRN)- Director of Adults with Down Syndrome Specialty Clinic, University of Kansas Medical Center, Kansas City, Kansas. She completed her dissertation in 2006 and the topic was adults with Down syndrome. In 2011 she started a primary care clinic for adults with Down syndrome within the Department of Family Medicine at the University of Kansas. It is a different model in that it has a nurse practitioner as the only healthcare provider. Peterson has been involved in the care of people with Down syndrome for most of her professional career. Her clinic is growing steadily and she enjoys this population so much.

# Medical Care Guidelines for Adults with DS

## *The Authors*



**Carl Tyler** (MD, MSc, ABFP, CAQ-Geriatrics)- Director of the Developmental Disabilities - Practice-Based Research Network (DD-PBRN), Practitioner at the Cleveland Clinic, Specializes in geriatrics and adults with I/DD, Cleveland, Ohio. His life work has been devoted to improving the health and health care of persons with developmental disabilities through clinical care, research, and professional and community education. His clinical training in psychiatry, family medicine, and geriatric medicine provided a rich clinical foundation and framework to understand the difficulties and complexities in providing health care to this population.



**Terry Harville**, MD, PhD, D(ABMLI), D(ABHI) is Professor of Pathology and Internal Medicine, Medical Director for the Histocompatibility Laboratory and Medical Director of the Immunogenetics and Transplantation Laboratory at the University of Arkansas for Medical Sciences (UAMS). He is also the Medical Director of the UAMS ABG Laboratory. He is a specialist in Pediatric Allergy, Asthma, Immunology, Rheumatology, Autoimmunity, Hematopoietic Stem cell Transplantation for Immunodeficiencies, and Organ Transplantation Immunology. He has a PhD in biochemistry and molecular biology from the University of Florida (UF) and an MD degree from the UF College of Medicine. Following residency in pediatrics and fellowship training in pediatric immunology, rheumatology, and transplantation biology at UF, he joined the faculty of Pediatric Allergy and Immunology at Duke University in Durham, NC, before matriculating to UAMS.

### **...Plus the ECRI Institute Team!**

- **Dr. Joann Fontanarosa**, PhD Senior Analyst, Co-Investigator ECRI Institute
- **Dr. Amy Tsou**, MD, MSc Principle Investigator, ECRI Institute
- **Allison Gross**, MS, MLS, Medical Librarian
- **Gina Giradi**, MS Project Coordinator
- **Janice Kaczmarek**, MS, Project Manager
- **Karen Schoelles**, MD, SM FACP EPC Director



# Medical Care Guidelines for Adults with DS

## *The Co-Authors*



**Peter Bulova (MD)**- Director of University of Pittsburgh Adult Down Syndrome Center, Pittsburgh, Pennsylvania. Dr. Bulova is an Associate Professor of General Internal Medicine. He completed his undergraduate training at Brown University. His medical school, internal medicine residency and chief residency took place at the University of Pittsburgh. He is on the Executive Board of the Down Syndrome Medical interest Group. He is clinically involved in research on Alzheimer's disease in adults with Down syndrome. He is on the NIH National Down Syndrome Registry Data Access Review Committee and is an ad hoc reviewer for several journals including Journal of Intellectual Disability Research, and the American Journal on Intellectual and Developmental Disabilities.



**Bryn Gelaro (AM, LSW)**- Director of Adult Initiatives and Special Projects, Global Down Syndrome Foundation, Denver, Colorado. Bryn Gelaro, a social worker with a passion for behavioral health of adults with Down syndrome, was a consultant for the Global Down Syndrome Foundation from 2015 to 2017 before becoming the Director of Adult Initiatives and Special Projects. In addition to her work with Global, she previously consulted for the former Denver Adult Down Syndrome Clinic. Bryn earned her Bachelors of Science in Psychology from the Pennsylvania State University in 2012 and a Masters (AM) in social work from the University of Chicago's School of Social Service and Administration in 2015. She completed her Master's graduate field training at the Chicago Adult Down Syndrome Clinic.



**Michael Wells (BS)**- Research Coordinator for the Developmental Disabilities - Practice-Based Research Network (DD-PBRN), Cleveland, Ohio. The DD-PBRN is a community-based participatory research network comprised uniquely of health care professionals, nurses, residential service providers, advocates and self-advocates. As Research Coordinator, Michael facilitates communication amongst these stakeholders, partner groups and prospective contacts or partners for future work. A major part of the coordinator role is dissemination of past, present and future work through academic lecture and poster presentations locally, regionally and nationally.

# Medical Care Guidelines for Adults with DS

## *Expert Volunteer Committee Members*

- ❖ Jarrett Barnhill (MD DFAPA FAACAP) Professor and Director, UNC Developmental Neuropharmacology Clinic
- ❖ Donald Bodenner (MD, PhD) Professor, Department of Geriatrics, University of Arkansas for Medical Sciences, Director of Thyroid Center and Chief of Endocrine Oncology
- ❖ Paul Camarata (MD) Professor and Chairman, Vascular & Skull Base Neurosurgery, University of Kansas Medical Center
- ❖ Kamala Gullapalli Cotts (MD) Associate Professor of Medicine, Director, Adult Developmental Disabilities Clinic, Section of General Medicine, Department of Medicine, The University of Chicago
- ❖ Robert Eckel (MD) Charles A. Boettcher II Endowed Chair in Atherosclerosis Professor of Medicine - Division of Endocrinology, Metabolism and Diabetes, and Cardiology, Professor of Physiology and Biophysics, Director of Lipid Clinic, University of Colorado Hospital
- ❖ Anna Esbensen (PhD) Associate Professor of Pediatrics, Cincinnati Children's Hospital
- ❖ James Hunt (MD) Assistant Professor of Anesthesiology, UAMS COM, Division of Pediatric Anesthesiology and Pain Medicine, Arkansas Children's Hospital
- ❖ Seth Keller (MD) Co-Chair, National Task Group on Intellectual Disabilities and Dementia Practices Chair, Special Interest Group Adult IDD, American Academy of Neurology Past President, American Academy of Developmental Medicine and Dentistry



# Medical Care Guidelines for Adults with DS

## *Expert Volunteer Committee Members*

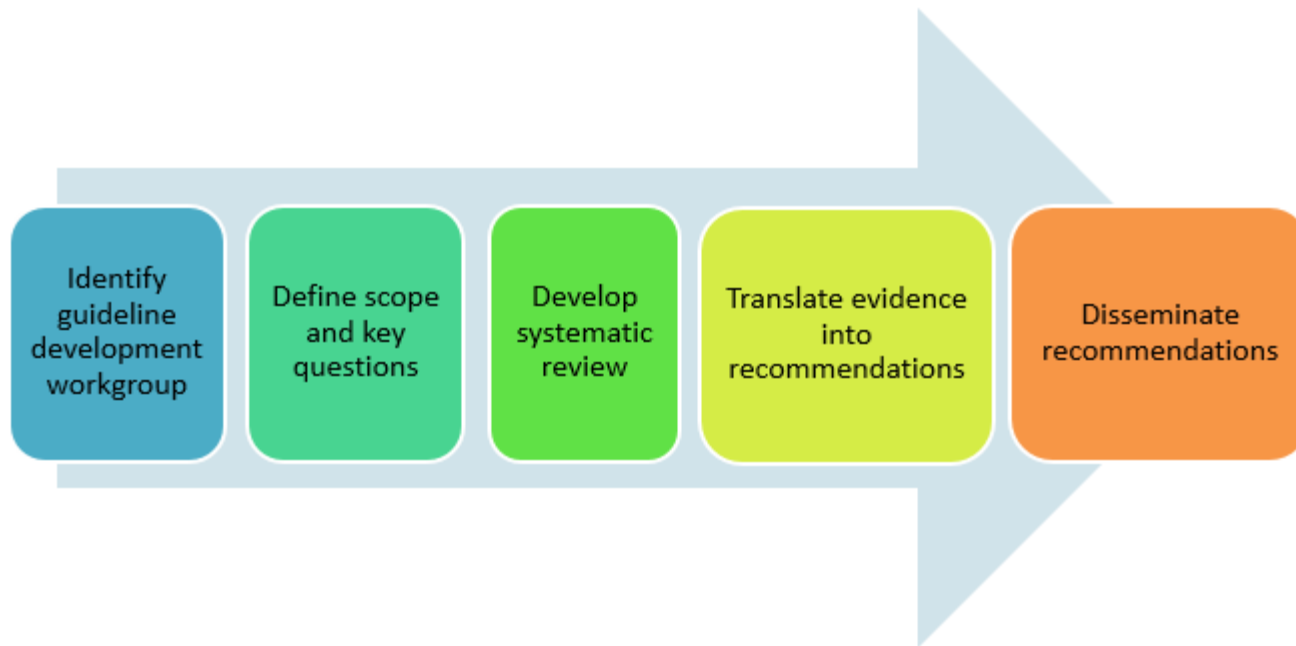
- ❖ Judy Kim (MD) Assistant Professor, Department of Medicine, Transition Medicine, Baylor College of Medicine
- ❖ Ira Lott (MD) Pediatric Neurologist, Emeritus Professor, University of California, Irvine and CHOC Children's Hospital
- ❖ Michael McDermott (MD) Professor of Medicine and Clinical Pharmacy, Division of Endocrinology, Diabetes Metabolism, University of Colorado School of Medicine
- ❖ Joan Medlen (MEd, RD, LD) Nutrition Counseling and Registered Dietitian
- ❖ Micol Rothman (MD) Associate Professor of Medicine, Director of the Metabolic Bone Program, Department of Medicine, Division of Endocrinology, University of Colorado Denver
- ❖ Stephanie Santoro (MD) Director of Quality Improvement Research, Down Syndrome Program, MassGeneral Hospital
- ❖ Mary Stephens (MD, MPH) Family Medicine and Jefferson Continuing Care Program, Jefferson Health and Down syndrome Consult Program, Christiana Care Center for Special Health Care Needs
- ❖ Elizabeth Yeung (MD) Associate Professor of Clinical Practice, Pediatric and Adult Congenital Cardiology, University of Colorado School of Medicine

# Medical Care Guidelines for Adults with DS

## *The Process*

### Overview of guideline development process

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# Medical Care Guidelines for Adults with DS

## *Topics to be Covered*

- ❖ **The initial update of the adult medical care guidelines will address the following 9 health areas as they pertain to adults with DS:**
  1. Cardiac Metabolism
  2. Muscular Skeletal
  3. Thyroid-Immunology
  4. Bone Density
  5. Metabolic Disorders
  6. Behavior/Mental Health
  7. Dementia
  8. GI-Immune-Special Diets
  9. Obesity-Lifestyle Activity-Special Diets

# Medical Care Guidelines for Adults with DS:

## *The Process*

### ❖ **PICOTS**

- Key questions directly inform the literature review and the types of recondition statements that can be created
- PICTOS is the evidence-based framework each key question must follow

### ❖ **GRADE**

- Process for translating evidence into recommendations

### ❖ **Ensure rigorous, evidence based, and transparent**

### ❖ **Standardizing quality of evidence**

- Used to determine direction & strength of recommendation
- Plus review of 4 domains

### ❖ **Endorsed by ACOG, CDC, AAFP, HICPAC**

# Medical Care Guidelines for Adults with DS

## *Addressing Research Gaps*

### ❖ **Future research needs**

- Each topic included a section on future research needs
- Gaps to be addressed before second iteration—“Road map”
- Improve strength of recommendations and increase knowledge base for better guidelines in the future

### ❖ **Example gaps identified include:**

1. Genetic risk factors for Type 2 diabetes
2. Energy metabolism in Down syndrome related to health and fitness
3. Bone mineral and fracture models specific to Down syndrome



# Our Accomplishments thus far...

- ✓ Recruited expert guideline creation group (ECRI)
- ✓ Recruited 11 expert DS clinicians as authors and co-authors
- ✓ Collectively wrote and finalized 20 critical questions—key indicators of health for adults with Down syndrome
- ✓ Formed 9 separate committees organized by medical content area
- ✓ Each committee recruited 1-3 additional volunteers for a total of 16 additional volunteers
- ✓ ECRI combed through thousands of studies to locate only the best possible evidence
- ✓ Evidence was given to committees who have been reviewing and are now coming to conclusions
- ✓ Authors met in person and agreed upon final recommendation statements
- ✓ Wrote the narratives that will become the physical guideline manuscript
- ✓ Facilitated family, self-advocate and provider focus groups & peer review!

# Focus Group & Peer Review

## ❖ Overachieving!

- Not required
- Some guidelines do a focus group or a peer review—we did both!

## ❖ Focus group

- 10 participants is recommended
- With help of NDSC and multiple local Down syndrome organizations, we recruited and retained 27 family members/caregivers and 7 self advocates of diverse age, location, gender, and race
- 7 day online discussion forum format

## ❖ Peer Review

- 5 participants is recommended
- We recruited 7 medical professional peer reviewers!

## ❖ Important needs identified:

- Glossary of medical terminology
- Patient/Family version of the guidelines
- Patient/Provider Toolkits to make the guideline easy to use



# Medical Care Guidelines for Adults with DS

## Global's Commitment

### ❖ GLOBAL IS COMMITTED TO

- Working with medical and health professionals and with national health organizations
- Creating guidelines and a useful checklist “tool”
- Translating the guidelines and checklist tool into several languages
- Updating the guidelines every 5 years
- Working to get the guidelines into major journals
- Ensuring the guidelines are available free and easily to families, providers and Down syndrome organizations

### ❖ ESTIMATED COST & TIMELINE

- \$450,000
- 24 months to create, vet and publish

# Medical Care Guidelines for Adults with DS Global's Commitment



## GLOBAL AIMS TO COVER ½ THE COST

- A HUGE thank you to all of YOU!
- Already raised \$130K from Local DS organizations!
- **Currently \$50K from our goal!**
- 50 local Down syndrome organizations and over 100 individuals have supported so far!

### THANK YOU TO OUR GENEROUS COMMUNITY SUPPORTERS!

Investing in Global's Medical Care Guidelines for Adults with Down syndrome

#### KEY COLLABORATOR

The Ritter Family

#### BENEFACTOR

Down Syndrome Association of Greater Richmond  
Down Syndrome Association of Minnesota  
Rocky Mountain Down Syndrome Association

#### KEY SUPPORTER

Anna & John J. Sie Center for Down Syndrome  
Down Syndrome Association of Central Ohio  
Down Syndrome Association of Jacksonville  
Down Syndrome Association of Orange County  
Down Syndrome Connection of the Bay Area  
Down Syndrome Guild of Dallas  
Linda Crnic Institute for Down Syndrome  
National Down Syndrome Congress

#### SUPPORTER

Broward Gold Coast Down Syndrome Organization  
ChapTer 21  
Down Syndrome Affiliates in Action  
Down Syndrome Alabama  
Down Syndrome Alliance of the Midlands  
Down Syndrome Association of Acadiana  
Down Syndrome Association of Central Florida  
Down Syndrome Association of Connecticut  
Down Syndrome Association for Families of Nebraska  
Down Syndrome Association of Greater Cincinnati  
Down Syndrome Association of Greater St. Louis  
Down Syndrome Association of Middle Tennessee  
Down Syndrome Association of Northern Virginia  
Fun Coast Down Syndrome Association  
The Family of Rya Gracyn Pierce

#### FRIEND

Chattanooga Down Syndrome Society  
Club 21 Learning and Resource Center  
Designer Genes of North Dakota  
Down Syndrome Association of Central California  
Down Syndrome Association of Central Kentucky  
Down Syndrome Association of Central Oklahoma  
Down Syndrome Association of Central Texas  
Down Syndrome Association of the Breckenridge Valley  
Down Syndrome Association of Wisconsin, Inc.  
Down Syndrome Family Connection  
Down Syndrome Foundation of Southeastern New Mexico  
Eastern Idaho Down Syndrome Family Connect  
East Texas Down Syndrome Group  
Families Exploring Down Syndrome of Broward  
International Music Down Syndrome Association  
Kern Down Syndrome Network  
Red River Valley Down Syndrome Society  
Rio Grande Valley Down Syndrome Association  
Surgcon Collaborative Law  
S.M.I.L.E. on Down Syndrome  
Southern Arizona Network for Down Syndrome  
The Up Side of Downs of Northeast Ohio  
Wisconsin Upside Down

*Adults With Down Syndrome Medical  
Care Guideline Status Update*

# Medical Care Guidelines for Adults with DS

## What the Guidelines Would Look Like



### Global Down Syndrome Foundation's medical care guidelines for adults with Down syndrome

Contributor/Author 1<sup>a</sup>, Contributor/Author 2<sup>a</sup>, Contributor/Author 3<sup>b,c</sup>,  
Contributor/Author 4<sup>d</sup>, Contributor/Author 5<sup>e</sup>, Contributor/Author 6<sup>f</sup>,  
Contributor/Author 7<sup>g</sup>;

*Organization Name, State, Country of Contributor/Author 1<sup>a</sup>*

*Organization Name, State, Country of Contributor/Author 2<sup>a</sup>*

*Organization Name, State, Country of Contributor/Author 3<sup>b,c</sup>*

*Organization Name, State, Country of Contributor/Author 4<sup>d</sup>*

*Organization Name, State, Country of Contributor/Author 5<sup>e</sup>*

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**Abstract** Guideline abstract will go here.

**Keywords:** Down syndrome, Developmental Disability, Alzheimer's disease, Assessment, Prevention, Sleep Apnea, Cognitive Impairment, Screening



# Medical Care Guidelines for Adults with DS

## What the Guidelines Would Look Like

### Prepared by:

#### Guideline Working Group

<b>Behavior-Dementia Volunteer Committee</b>	<b>Behavior-Mental Health Volunteer Committee</b>	<b>Bone Density Volunteer Committee</b>
Lead Authors: Capone, G., Chicoine, B., & McGuire, D. Co-Author: Gelaro, B. Volunteers: Esbensen, A., & Barnhill, J.	Lead Authors: Capone, G., & McGuire, D. Co-Author: Gelaro, B. Volunteers: Keller, S., & Lott, I.	Lead Authors: McKelvey, K., & Tyler, C. Co-Author: Wells, M. Volunteer: Rothman, M.
<b>Cardiac-Metabolic Volunteer Committee</b>	<b>GI-Immune-Special Diets Volunteer Committee</b>	<b>Metabolic Disorders Volunteer Committee</b>
Lead Authors: Chicoine, B., & Martin, B. Co-Author: Bulova, P. Volunteers: Eckel, R., & Yeung, E.	Lead Authors: McKelvey, K., Tyler, C., & Harville, T. Co-Author: Wells, M.	Lead Authors: Chicoine, B., & Peterson, M. Volunteers: Santoro, S., & Stephens, M.
<b>Muscular-Skeletal Volunteer Committee</b>	<b>Obesity-Lifestyle-Activity-Special Diets Volunteer Committee</b>	<b>Thyroid Volunteer Committee</b>
Lead Authors: Martin, B., & Peterson, M. Volunteers: Hunt, J., Camarata, P., & Stephens, M.	Lead Authors: Capone, G., & Peterson, M. Co-Author: Bulova, P. Volunteers: Kim, J., Medlen, J., & Cotts, K.	Lead Authors: Martin, B., & McKelvey, K. Volunteers: Bodenner, D., & McDermott, M.

#### The ECRI Institute

Amy Tsou (Principle Investigator)  
Joann Fontanarosa (Co P.I.)  
Allison Gross (Medical Librarian)  
Gina Giradi (Project Coordinator)  
Janice Kaczmarek (Project Manager)  
Karen Schoelles (EPS Director)

# Medical Care Guidelines for Adults with DS

## What the Guidelines Would Look Like

### With support from:

#### Key Collaborators

The Ritter Family

#### Benefactors

Down Syndrome Association of Greater Richmond  
Down Syndrome Association of Minnesota  
Rocky Mountain Down Syndrome Association

#### Key Supporters

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Down Syndrome Association of Orange County  
Down Syndrome Connection of the Bay Area  
Down Syndrome Guild of Dallas  
Linda Crnic Institute  
National Down Syndrome Congress

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Down Syndrome Association of Middle Tennessee	Fun Coast Down Syndrome Association
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Down Syndrome Association for Families of Nebraska	

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Down Syndrome Association of Central Oklahoma	Down Syndrome Family Connection
Down Syndrome Association of Central Texas	Down Syndrome Foundation of Southeastern New Mexico
Down Syndrome Association of the Brazos Valley	East Texas Down Syndrome Group
Kern Down Syndrome Network	Eastern Idaho Down Syndrome Family Connect
Rio Grande Valley Down Syndrome Association	Families Exploring Down Syndrome of Brevard
Sampson Collaborative Law	International Mosaic Down Syndrome Association
Southern Arizona Network for Down Syndrome	Red River Valley Down Syndrome Society
The UpSide of Downs of Northeast Ohio	S.M.I.L.E. on Down Syndrome
Down Syndrome Association of Delaware	Wisconsin Upside Down

# Medical Care Guidelines for Adults with DS

## What the Guidelines Would Look Like



### Example Health Care Guideline Appointment Tool for Adults with Down Syndrome (2019)


February 2019

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# Pediatric Medical Care Record Sheet

Courtesy of Anna & John J Sie Center for Down Syndrome



Children's Hospital Colorado

Anna and John J. Sie Center for Down Syndrome

## Down Syndrome Healthcare Guidelines (2011 Revision) Record Sheet\*

	Birth	6 mo	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20														
Genetic Counseling <sup>1</sup> , Karyotype																																				
Parent Group Info and Support																																				
CBC to R/O transient myeloproliferative disorder, polycythemia		Parent-to-parent contact, support groups, current books and pamphlets																																		
Swallowing assessment if feeding problems or aspiration																																				
Hemoglobin			Hemoglobin annually beginning at 1 year old. If Hg<11, do (a) CRP and ferritin, or (b) Reticulocyte Hemoglobin Content (CHr). If possible risk for iron deficiency, do (a) or (b) regardless.																																	
23-valent pneumococcal vaccine <sup>2</sup>																																				
Cardiology	Echo <sup>4</sup>															Screen for acquired mitral or aortic valvular disease																				
Audiological Evaluation	ABR or OAE	Every 6 months till 3 years of age. Annually thereafter.																																		
Ophthalmologic Evaluation	Red Reflex	Optho Appt	Annual ophthalmology appt					Q2 Ophthalmology appointment							Q3 Ophthalmology appointment																					
Celiac Disease Screening						(Only test if signs and symptoms present)																														
Thyroid – TSH, T4	State Screen	Test	Test	Test TSH and T4 annually																																
Neck X-ray (AAI) <sup>3</sup>					✓ <sup>3</sup>																															
Dental Exam			Annual Dental Exams. Reassure parents that delayed or irregular eruption, hypodontia are common.																																	
Sleep Study by age 4 years	Done prior to 4 years of age																																			
Early Intervention																																				
Childhood						Discuss self-help, ADHD, OCD, wandering off, transition to middle school																														
Puberty																Discuss physical and psychosocial changes through puberty, need for gynecologic care (pelvic exams) in pubescent female																				
Facilitate transition																Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group home, work settings																				
Sexual development and behaviors																Discuss Contraception, STDs, recurrence risk for offspring																				
Preventive care	Annually monitor for signs and symptoms of constipation, OSA, and aspiration.																																			

1. Discuss Recurrence Rate of future pregnancies with parents

2. 23-valent pneumococcal vaccine if chronic or pulmonary disease.

3. AAI: See AAP Guidelines page 399 - X-rays only if myopathic signs or symptoms \* Peds 2011 ;128 :393-406 Chart by Sie Center for Down Syndrome

4. Follow up to be determined by Cardiologist

Updated 09/2013

Confidential

# Multi-year Plan

## ❖ 2020 Publish first version with 9 areas

- Mostly suggestions and recommendations versus guidelines
- Work with other organizations to ensure broad, free, and unedited distribution

## ❖ 2020-2024

- Translate the guidelines into several languages and work with international medical organizations on the distribution
- Work with other organizations to ensure broad distribution
- Work with clinical researchers to help bolster evidence in the 9 areas and 1-2 new areas
- Help to fundraise in this regard as necessary

## ❖ 2023 Start Planning for 2<sup>nd</sup> version with a 5 year update





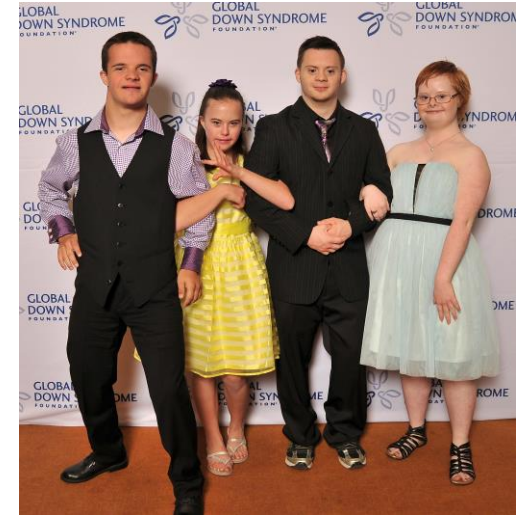
# Critical NEXT STEPS

- ❖ Mapping out research to address “gaps” in evidence
- ❖ Finalizing the “checklist” tools and other Patient/provider tools based off recommendations
- ❖ Submitting to medical journals
  - Open Access
  - Online & print
  - Reputable
- ❖ Strategizing distribution & dissemination



# How You Can Help Elongate and Improve Quality of Life

- ❖ **Donate!** Help us raise the final \$50K...
- ❖ Help us to distribute the guidelines once they are published
- ❖ Hold workshops for parents and professional around the guidelines
- ❖ Help us make the guidelines financially sustainable in future years
- ❖ Help us fund research between now and the next guidelines
- ❖ Help us advocate in Washington DC for UNDERWRITTEN medical centers of excellence



# THANK YOU TO OUR LEGACY SUPPORTERS!

## Key Collaborators

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The Ritter Family

## Benefactors

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Down Syndrome Association of Greater Richmond  
Down Syndrome Association of Minnesota  
Rocky Mountain Down Syndrome Association

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Down Syndrome Association of Orange County  
Down Syndrome Connection of the Bay Area  
Down Syndrome Guild of Dallas  
Linda Crnic Institute  
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# Q + A

