



GLOBAL
DOWN SYNDROME FOUNDATION®

Medical Guidelines For Adults with Down Syndrome

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Global Research and Medical Care Roundtable
June 25, 2020

Global Medical Care Guidelines for Adults with DS

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Global Medical Care Guidelines for Adults with DS

The Co-Authors



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...Plus Global and the ECRI Institute Team!

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Medical Care Guidelines for Adults with DS

Project Timeline

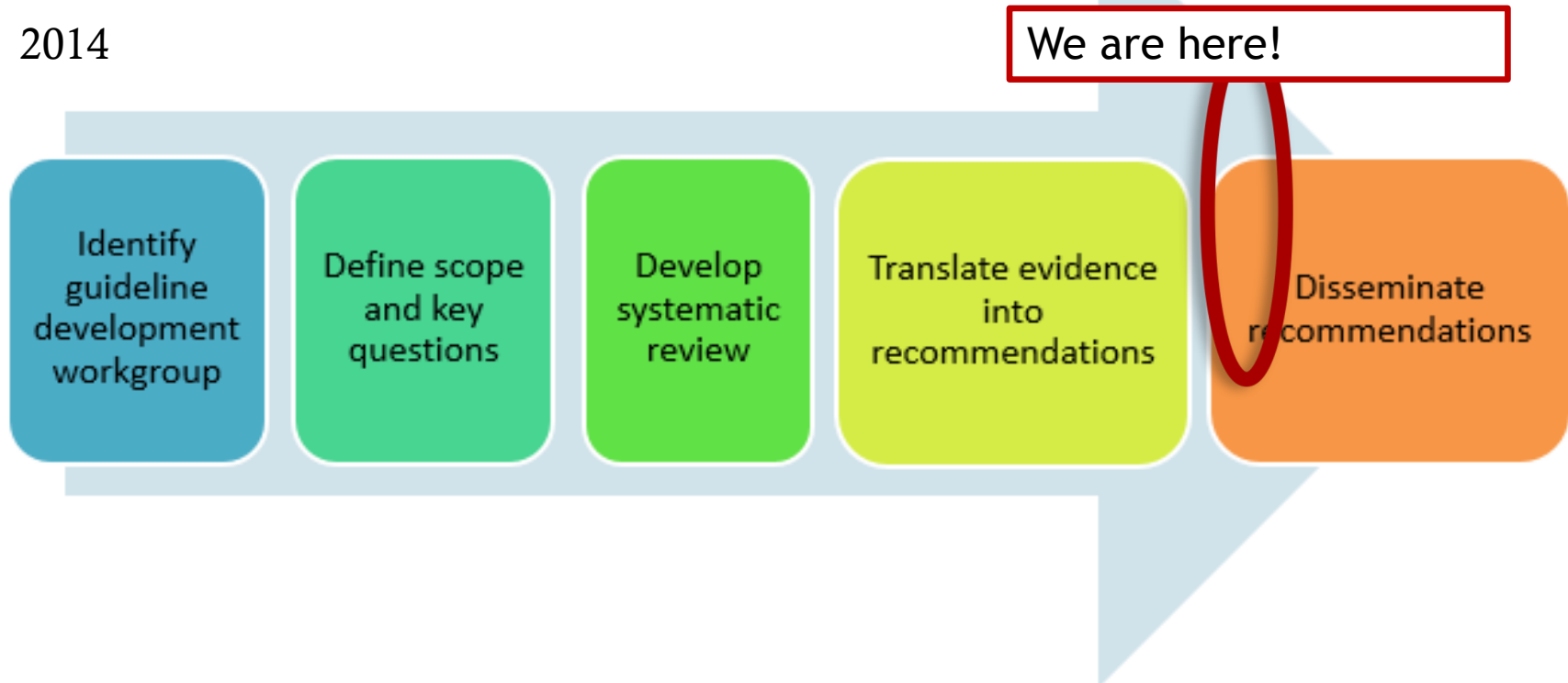
- ❖ **2014**—Identified guidelines as key project
- ❖ **2015**—DSMIG-USA Adult Workgroup identified key areas of research
- ❖ **2017-2018**—Global assembled 9 Down syndrome experts to join project and formed work group.
- ❖ **2018** Key topics agreed upon, literature review performed by ECRI
- ❖ **2019** In-person meeting to translate evidence into recommendations in Denver, CO
- ❖ **Today**

Global Medical Care Guidelines for Adults w/ DS

A 6 year process

Overview of guideline development process

2014

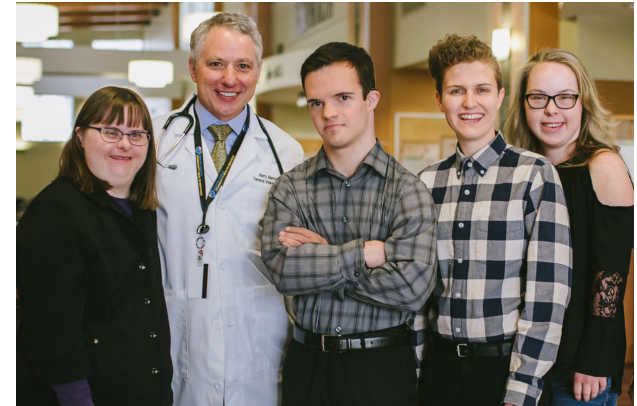


Guideline Questions

A Daunting Task

❖ 70 pertinent clinical questions were generated and triaged to nine major subjects

1. Behavior/Mental Health
2. Dementia
3. Diabetes
4. Cardiovascular Disease
5. Obesity
6. Atlantoaxial Instability
7. Osteoporosis
8. Thyroid
9. Celiac Disease



Questions & Qualifying Studies

A Daunting Task

11,295 possible
citations Identified



7,200 abstracts
reviewed



1,929 Articles
reviewed round 1



199 Articles
reviewed round 2



**20 Studies
Qualify**

❖ **The 70 questions were weaned to 26 key questions**

- Reviewed hundreds of thousands of publications and vetted the small qualifying work for best practices and evidence with Global and the authors

❖ **Of over 11,000 only 20 high quality studies!**

Evidence by Question

Our Desperate Need for Research

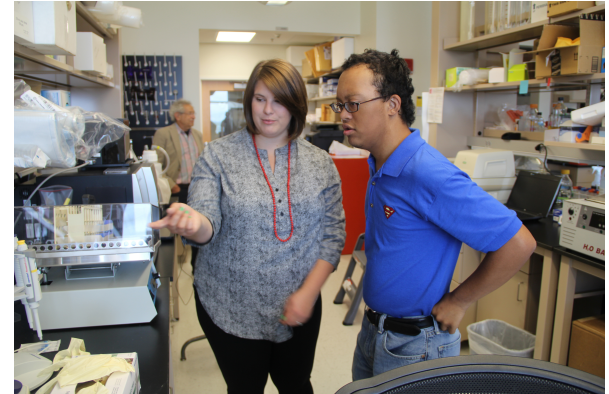
FOURTEEN have “No Evidence” – what does that mean?

Question #	Category Question	Evidence Found	Question #	Category Question	Evidence Found
1	Behavior/Mental Health	2 Low	14	Musculoskeletal	No Evidence
2	Behavior/Mental Health	No Evidence	15	Bone Density	6 Low
3	Dementia	1 High, 2 Moderate	16	Bone Density	No Evidence
4	Metabolic Disorders	2 High, 3 Moderate	17	Bone Density	2 Low
5	Metabolic Disorders	No Evidence	18	Bone Density	1 Moderate
6	Metabolic Disorders	No Evidence	19	Thyroid	1 High, 1 Moderate, 1 Low
7	Cardiac Metabolism	1 High, 1 Low	20	Thyroid	No Evidence
8	Cardiac Metabolism	1 Low	21	Thyroid	No Evidence
9	Cardiac Metabolism	No Evidence	22	Thyroid	No Evidence
10	Obesity-Lifestyle	2 Low	23	GI-Immune	2 Low
11	Obesity-Lifestyle	No Evidence	24	GI-Immune	No Evidence
12	Obesity-Lifestyle	No Evidence	25	GI-Immune	No Evidence
13	Musculoskeletal	2 Moderate	26	GI-Immune	No Evidence

What Will be Included in the Guidelines

❖ Key areas addressed:

- How common is a condition?
- How is it diagnosed?
- How do we treat?



❖ We will provide guidance around:

- Frequency of screening
- Assessment tools to use (or not use)
- Updates to existing guidelines for people w/o DS (*Ex: American Diabetes Association Guidelines*)

What Will be Included in the Guidelines

Recommendations & SOGP

❖ Final Product:

- **Statements of Good Practice: 4**
 - Obesity
 - GI-Immune
 - Behavior (2)
- **Recommendation Statements: 14**
- **1 “Strong” Recommendation**
 - Dementia
- **12 “Weak” Recommendations**
 - Still provide useful guidance; they are based on less evidence and more expert insight
- **1 “No” Recommendation**



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 ¹ , 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 ²																						
Cardiology	Echo ⁴														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) ³					✓ ³																	
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.

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

Global's Multi-year Plan for the Guidelines

We are in this for the long haul...

❖ Future

- Oct/Nov 2020 - publish
- Work with Global Members & Guideline Supporters to ensure broad, free, and unedited distribution



Global's Multi-year Plan for the Guidelines

We are in this for the long haul - we hope you are too



❖ How You Can Help:

- Use the guidelines!!!
- Talk to your provider about the guidelines!!!
- Donate to support ongoing work
- Encourage local researchers and clinicians to participate in appropriate research
- Participate in advocacy for adult care and funding



This is an iterative process

- ❖ 2026 Publish Updated guidelines and new topics

Global Medical Care Guidelines for Adults w/ DS

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

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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



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