Global Medical Care Guidelines for Adults with DS

The Authors

Dr. Peter Bulova
Univ Pittsburg, PA

Dr. George Capone
Kennedy Krieger, MD

Dr. Dennis McGuire
Chicago Adult DS Clinic, IL

Dr. Moya Peterson
Kansas Adult DS Clinic, KS

Dr. Brian Chicoine
Chicago Adult DS Clinic, IL

Dr. Barry Martin
UCH, CO

Dr. Kent McKelvey
UAMS, AR

Dr. Carl Tyler
Cleveland Clinic, OH

Dr. Terry Harville
UAMS, AR
Global Medical Care Guidelines for Adults with DS

The Co-Authors

Bryn Gelaro, LSW
Denver, CO

Michael Wells, BS
DD-PBRN Cleveland, OH

...Plus Global and the ECRI Institute Team!

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

2020 Global Roundtable
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**
A 6 year process

Overview of guideline development process

Identify guideline development workgroup
Define scope and key questions
Develop systematic review
Translate evidence into recommendations
Disseminate recommendations

2014

We are here!
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

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

<table>
<thead>
<tr>
<th>Question #</th>
<th>Category Question</th>
<th>Evidence Found</th>
<th>Question #</th>
<th>Category Question</th>
<th>Evidence Found</th>
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</thead>
<tbody>
<tr>
<td>1</td>
<td>Behavior/Mental Health</td>
<td>2 Low</td>
<td>14</td>
<td>Musculoskeletal</td>
<td>No Evidence</td>
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<tr>
<td>2</td>
<td>Behavior/Mental Health</td>
<td>No Evidence</td>
<td>15</td>
<td>Bone Density</td>
<td>6 Low</td>
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<td>3</td>
<td>Dementia</td>
<td>1 High, 2 Moderate</td>
<td>16</td>
<td>Bone Density</td>
<td>No Evidence</td>
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<tr>
<td>4</td>
<td>Metabolic Disorders</td>
<td>2 High, 3 Moderate</td>
<td>17</td>
<td>Bone Density</td>
<td>2 Low</td>
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<tr>
<td>5</td>
<td>Metabolic Disorders</td>
<td>No Evidence</td>
<td>18</td>
<td>Bone Density</td>
<td>1 Moderate</td>
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<tr>
<td>6</td>
<td>Metabolic Disorders</td>
<td>No Evidence</td>
<td>19</td>
<td>Thyroid</td>
<td>1 High, 1 Moderate, 1 Low</td>
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<tr>
<td>7</td>
<td>Cardiac Metabolism</td>
<td>1 High, 1 Low</td>
<td>20</td>
<td>Thyroid</td>
<td>No Evidence</td>
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<tr>
<td>8</td>
<td>Cardiac Metabolism</td>
<td>1 Low</td>
<td>21</td>
<td>Thyroid</td>
<td>No Evidence</td>
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<tr>
<td>9</td>
<td>Cardiac Metabolism</td>
<td>No Evidence</td>
<td>22</td>
<td>Thyroid</td>
<td>No Evidence</td>
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<tr>
<td>10</td>
<td>Obesity-Lifestyle</td>
<td>2 Low</td>
<td>23</td>
<td>GI-Immune</td>
<td>2 Low</td>
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<tr>
<td>11</td>
<td>Obesity-Lifestyle</td>
<td>No Evidence</td>
<td>24</td>
<td>GI-Immune</td>
<td>No Evidence</td>
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<td>12</td>
<td>Obesity-Lifestyle</td>
<td>No Evidence</td>
<td>25</td>
<td>GI-Immune</td>
<td>No Evidence</td>
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<td>13</td>
<td>Musculoskeletal</td>
<td>2 Moderate</td>
<td>26</td>
<td>GI-Immune</td>
<td>No Evidence</td>
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</tbody>
</table>
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)
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
# Down Syndrome Healthcare Guidelines (2011 Revision) Record Sheet

<table>
<thead>
<tr>
<th>Genetic Counseling, Karyotype</th>
<th>Birth</th>
<th>1</th>
<th>2</th>
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<tbody>
<tr>
<td>CBC to R/O transient myeloproliferative disorder, polycythemia</td>
<td>Parent-to-parent contact, support groups, current books and pamphlets</td>
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<td>Swallowing assessment if feeding problems or aspiration</td>
<td>Hemoglobin annually beginning at 1 year old. If Hg&lt;11, do (a) CRP and ferritin, or (b) Reticulocyte Hemoglobin Content (CHR). If possible risk for iron deficiency, do (a) or (b) regardless.</td>
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<tr>
<td>Hemoglobin</td>
<td>23-valent pneumococcal vaccine</td>
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<tr>
<td>Cardiology</td>
<td>Echo&lt;sup&gt;1&lt;/sup&gt;</td>
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<td>Cardiology</td>
<td>Every 6 months till 3 years of age. Annually thereafter.</td>
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<td>Audiolgical Evaluation</td>
<td>ABR or OAE</td>
<td>Screen for acquired mitral or aortic valvular disease</td>
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<td>Ophthalmologic Evaluation</td>
<td>Red Reflex</td>
<td>Annual ophthalmology appointment</td>
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<td>Q2 Ophthalmology appointment</td>
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<td>Q3 Ophthalmology appointment</td>
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<tr>
<td>Celiac Disease Screening</td>
<td>(Only test if signs and symptoms present)</td>
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<tr>
<td>Thyroid – TSH, T4</td>
<td>State Screen</td>
<td>Test</td>
<td>Test</td>
<td>Test</td>
<td>TSH and T4 annually</td>
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<td>Neck X-ray (AAR)&lt;sup&gt;2&lt;/sup&gt;</td>
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<tr>
<td>Dental Exam</td>
<td>Annual Dental Exams. Reassure parents that delayed or irregular eruption, hypodontia are common.</td>
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<td>Sleep Study by age 4 years</td>
<td>Done prior to 4 years of age</td>
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<td>Early Intervention</td>
<td>Discuss self-help, ADHD, ODD, wandering off, transition to middle school</td>
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<tr>
<td>Childhood</td>
<td>Puberty</td>
<td>Discuss physical and psychosocial changes through puberty, need for gynecologic care (pelvic exams) in pubescent female</td>
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<td>Facilitate transition</td>
<td>Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group home, work settings</td>
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<tr>
<td>Sexual development and behaviors</td>
<td>Sexual development and behaviors</td>
<td>Discuss Contraception, STDs, recurrence risk for offspring</td>
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<tr>
<td>Preventive care</td>
<td>Preventive care</td>
<td>Annually monitor for signs and symptoms of constipation, OSA, and aspiration.</td>
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1. Discuss Recurrence Rate of future pregnancies with parents
2. 23-valent pneumococcal vaccine if chronic or pulmonary disease.
3. AAR: See AAP Guidelines page 399 - X-rays only if myopathic signs or symptoms
4. Follow up to be determined by Cardiologist

* Peds 2011;128:399-406 Chart by Sie Center for Down Syndrome

Updated 09/2013

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**2020 Global Roundtable**

**Global Medical Care Guidelines**
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

2020 Global Roundtable
Global Medical Care Guidelines for Adults w/ DS

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 Guidelines For Adults with Down Syndrome

Kent McKelvey, MD
Rockefeller Chair in Clinical Genetics
Associate Professor in Medical Genetics and Family Medicine
University of Arkansas for Medical Sciences

Global Research and Medical Care Roundtable
June 25, 2020