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
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!
“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
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.
Currently list over 60 clinics across the US

- 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?
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!

[Diagram showing the flow from Research, through Medical Professional, Patients, and back to Medical Professional and Research]
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?

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...
Alzheimer’s Association recommendations for operationalizing the detection of cognitive impairment during the Medicare Annual Wellness Visit in a primary care setting

Cyndy B. Cordell, MD,1,2 Soo B. B. B. So,1,3 Malaz Boustanian, MD,1,4 Joshua Chodosh, MD,1,5 David Reuber, MD,1,6 Joe Verghese,1 William Thies,6 Leslie B. Fried,1,6 the Medicare Detection of Cognitive Impairment Workgroup

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2Department of Psychiatry and Behavioral Sciences, University of Washington School of Medicine, Seattle, WA, USA
3University Hospitals Plainview and Diagnostic Health Services, University of Washington School of Medicine, Seattle, WA, USA
4Indiana University Center for Aging Research, Indianapolis, IN, USA
5Regenstrief Institute, Inc., Indianapolis, IN, USA
6Department of Medicine, Indiana University School of Medicine, Indianapolis, IN, USA
7University of California, David Geffen School of Medicine, University of California-San Francisco, Los Angeles, CA, USA
8Department of Nursing, Albert Einstein College of Medicine, Bronx, NY, USA
9American Bar Association, Washington, DC, USA
10Alzheimer’s Association, Washington, DC, USA

Abstract

The Patient Protection and Affordable Care Act added a new Medicare benefit, the Annual Wellness Visit (AWV), effective January 1, 2011. The AWV requires an assessment to detect cognitive impairment. The Centers for Medicare and Medicaid Services (CMS) elected not to recommend a specific assessment tool because there is no single, universally accepted assessment that satisfies all needs in the detection of cognitive impairment. To provide primary care physicians with guidance on cognitive assessment during the AWV, and when referred or further testing is needed, the Alzheimer’s Association convened a group of experts to develop recommendations. The resulting Alzheimer’s Association Medicare Annual Wellness Visit Algorithms for Assessment of Cognition includes review of patient Health Risk Assessment (HRA) information, patient observation, structured queries during the AWV, and use of structured cognitive assessment tools for both patients and informants. Widespread implementation of this algorithm could be the first step in reducing the prevalence of undiagnosed or delayed dementia diagnosis, thus allowing for better healthcare management and more favorable outcomes for affected patients and their families and caregivers. © 2013 The Alzheimer’s Association. All rights reserved.

Keywords: Annual Wellness Visit, AWV, Cognitive Impairment, Assessment, Source, Dementia, Alzheimer’s disease, Medicare, Algorithm, Patient Protection and Affordable Care Act.

1. Introduction

The Patient Protection and Affordable Care Act of 2010 added a new Medicare benefit, the Annual Wellness Visit (AWV), effective January 1, 2011. The AWV includes routine measurements such as height, weight, and blood pressure; a review of medical and family history; an assessment to detect cognitive impairment; and establishment of a list of current medical providers, medications, and schedule for future preventive services. In addition, during the first AWV only, beneficiaries are to be screened for depression (DSM-5).
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 outreach/ed opportunities
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 perform 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.
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.
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.
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
Overview of guideline development process

1. Identify guideline development workgroup
2. Define scope and key questions
3. Develop systematic review
4. Translate evidence into recommendations
5. Disseminate recommendations
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
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
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
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
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!
Global Down Syndrome Foundation’s medical care guidelines for adults with Down syndrome

Contributor/Author 1*, Contributor/Author 2*, Contributor/Author 3*h, Contributor/Author 4*, Contributor/Author 5*, Contributor/Author 6*, Contributor/Author 7*

Organization Name, State, Country of Contributor/Author 1*
Organization Name, State, Country of Contributor/Author 2*
Organization Name, State, Country of Contributor/Author 3*h
Organization Name, State, Country of Contributor/Author 4*
Organization Name, State, Country of Contributor/Author 5*

Abstract
Guideline abstract will go here.

Keyword:
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

<table>
<thead>
<tr>
<th>Prepared by:</th>
<th>Guideline Working Group</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Behavior-Dementia</strong></td>
<td><strong>Behavior-Mental Health</strong></td>
</tr>
<tr>
<td>Volunteer Committee</td>
<td>Volunteer Committee</td>
</tr>
<tr>
<td>Lead Authors: Capone, G., Chacon, B., &amp; McGuire, D. Co-Author: Gelaro, B. Volunteers: Eibesheim, A., &amp; Barnhill, J.</td>
<td>Lead Authors: Capone, G., &amp; McGuire, D. Co-Author: Gelaro, B. Volunteers: Keller, S., &amp; Lott, L.</td>
</tr>
<tr>
<td><strong>Cardiac-Metabolic</strong></td>
<td><strong>GI-Immune-Special Diets</strong></td>
</tr>
<tr>
<td>Volunteer Committee</td>
<td>Volunteer Committee</td>
</tr>
<tr>
<td>Lead Authors: Chicoine, B., &amp; Martin, B. Co-Author: Bulova, P. Volunteers: Eckel, R., &amp; Young, E.</td>
<td>Lead Authors: McKelvey, K., Tyler, C., &amp; Harris, T. Co-Author: Wells, M.</td>
</tr>
<tr>
<td><strong>Muscular-Skeletal</strong></td>
<td><strong>Obesity-Lifestyle-Activity-Special Diets</strong></td>
</tr>
<tr>
<td>Volunteer Committee</td>
<td>Volunteer Committee</td>
</tr>
</tbody>
</table>

**The ECRI Institute**

- Amy Iscu (Principal Investigator)
- Joanna Fontanares (Co P.I.)
- Allison Gross (Medical Librarian)
- Gina Giradi (Project Coordinator)
- Janice Kaczmarek (Project Manager)
- Karen Schoelles (EPS Director)
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

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

Supporters

Down Syndrome Association of Northern Virginia
Down Syndrome Affiliates in Action
Down Syndrome Alliance of the Midlands
Down Syndrome Association of Middle Tennessee
Down Syndrome Alabama
Down Syndrome Association of Greater Cincinnati
Down Syndrome Association of Central Connecticut
Down Syndrome Association for Families of Nebraska

Friends

Chattanooga Down Syndrome Society
Club 21 Learning and Resource Center
Down Syndrome Association of Central Kentucky
Down Syndrome Association of Central Oklahoma
Down Syndrome Association of Central Texas
Down Syndrome Association of the Brazos Valley
Kern Down Syndrome Network
Rio Grande Valley Down Syndrome Association
San Pasqual Collaborative Learning
Southern Arizona Network for Down Syndrome
The Upside of Downs of Northeast Ohio
Down Syndrome Association of Delaware

Designer Genes of North Dakota
Down Syndrome Association of Central California
Down Syndrome Association of Wisconsin, Inc.
Down Syndrome Family Connection
Down Syndrome Foundation of Southeastern New Mexico
East Texas Down Syndrome Group
Eastern Idaho Down Syndrome Family Connection
Families Exploring Down Syndrome of Brevard
International Mosaic Down Syndrome Association
Red River Valley Down Syndrome Society
S.M.I.L.E. on Down Syndrome
Wisconsin Upside Down
# Example Health Care Guideline Appointment Tool for Adults with Down Syndrome (2019)

<table>
<thead>
<tr>
<th>Month</th>
<th>Year</th>
<th>Task Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>January</td>
<td>2019</td>
<td>Health Assessment</td>
</tr>
<tr>
<td>February</td>
<td>2019</td>
<td>Genetic Counseling</td>
</tr>
<tr>
<td>March</td>
<td>2019</td>
<td>Cardiac Screen</td>
</tr>
<tr>
<td>April</td>
<td>2019</td>
<td>Vision Evaluation</td>
</tr>
<tr>
<td>May</td>
<td>2019</td>
<td>Audiological Testing</td>
</tr>
<tr>
<td>June</td>
<td>2019</td>
<td>Dental Check-Up</td>
</tr>
<tr>
<td>July</td>
<td>2019</td>
<td>Nutritional Assessment</td>
</tr>
<tr>
<td>August</td>
<td>2019</td>
<td>Behavioral Therapy</td>
</tr>
<tr>
<td>September</td>
<td>2019</td>
<td>Occupational Therapy</td>
</tr>
<tr>
<td>October</td>
<td>2019</td>
<td>Respiratory Evaluation</td>
</tr>
<tr>
<td>November</td>
<td>2019</td>
<td>Gastrointestinal Consultation</td>
</tr>
<tr>
<td>December</td>
<td>2019</td>
<td>Dermatological Check</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Genetic Counseling, Karyotype</th>
<th>CBC to R/D transient myeloproliferative disorder, polycythemia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parent Group Info and Support</td>
<td>Parent-to-parent contact, support groups, current books and pamphlets</td>
</tr>
<tr>
<td>Swallowing assessment if feeding problems or aspiration</td>
<td>Hemoglobin annually beginning at 1 year old. If Hgb&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>
</tr>
<tr>
<td>23-valent pneumococcal vaccine</td>
<td>Cardiology</td>
</tr>
<tr>
<td>Echo</td>
<td>Screen for acquired mitral or aortic valvular disease</td>
</tr>
<tr>
<td>Audiological Evaluation</td>
<td>Cardiology</td>
</tr>
<tr>
<td>ABR or OAE</td>
<td>Cardiology</td>
</tr>
<tr>
<td>Every 6 months till 3 years of age. Annually thereafter.</td>
<td>Cardiology</td>
</tr>
<tr>
<td>Ophthalmologic Evaluation</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Red Reflex</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Ophtho Appt</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Annual ophthalmology appointment</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Q2 Ophthalmology appointment</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Q3 Ophthalmology appointment</td>
<td>Celiac Disease Screening</td>
</tr>
<tr>
<td>Celiac Disease Screening</td>
<td>Thyroid – TSH, T4</td>
</tr>
<tr>
<td>Screen</td>
<td>Thyroid – TSH, T4</td>
</tr>
<tr>
<td>Test</td>
<td>Thyroid – TSH, T4</td>
</tr>
<tr>
<td>Test</td>
<td>Thyroid – TSH, T4</td>
</tr>
<tr>
<td>Test TSH and T4 annually</td>
<td>Thyroid – TSH, T4</td>
</tr>
<tr>
<td>Neck X-ray (AAG)</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>✓²</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Annual Dental Exams. Reassure parents that delayed or irregular eruption, hypodontia are common.</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Sleep Study by age 4 years</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Done prior to 4 years of age</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Early Intervention</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Childhood</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Discuss self-help, ADHD, OCD, wandering off, transition to middle school</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Puberty</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Discuss physical and psychosocial changes through puberty, need for gynecologic care (pelvic exams) in pubescent female</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Facilitate transition</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group home, work settings</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Sexual development and behaviors</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Discuss Contraception, STDs, recurrence risk for offspring</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Preventive care</td>
<td>Dental Exam</td>
</tr>
<tr>
<td>Annually monitor for signs and symptoms of constipation, OSA, and aspiration.</td>
<td>Dental Exam</td>
</tr>
</tbody>
</table>

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1. Discuss Recurrence Rate of future pregnancies with parents  
2. 23-valent pneumococcal vaccine if chronic or pulmonary disease.  
3. AAG: See AAP Guidelines page 399 - X-rays only if myopathic signs or symptoms  
4. Follow up to be determined by Cardiologist

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Updated 08/2013

*Peds 2011;128:399-406 Chart by Sie Center for Down Syndrome
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 2nd 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!

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The Ritter Family

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Rocky Mountain Down Syndrome Association

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