Boston Children's Hospital
Down Syndrome Program

Important Updates in Pediatric Care & Research in Learning and Development for Children with Down Syndrome

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Boston Children's

Where the world comes for answers

Boston Children's Hospital
Down Syndrome Program

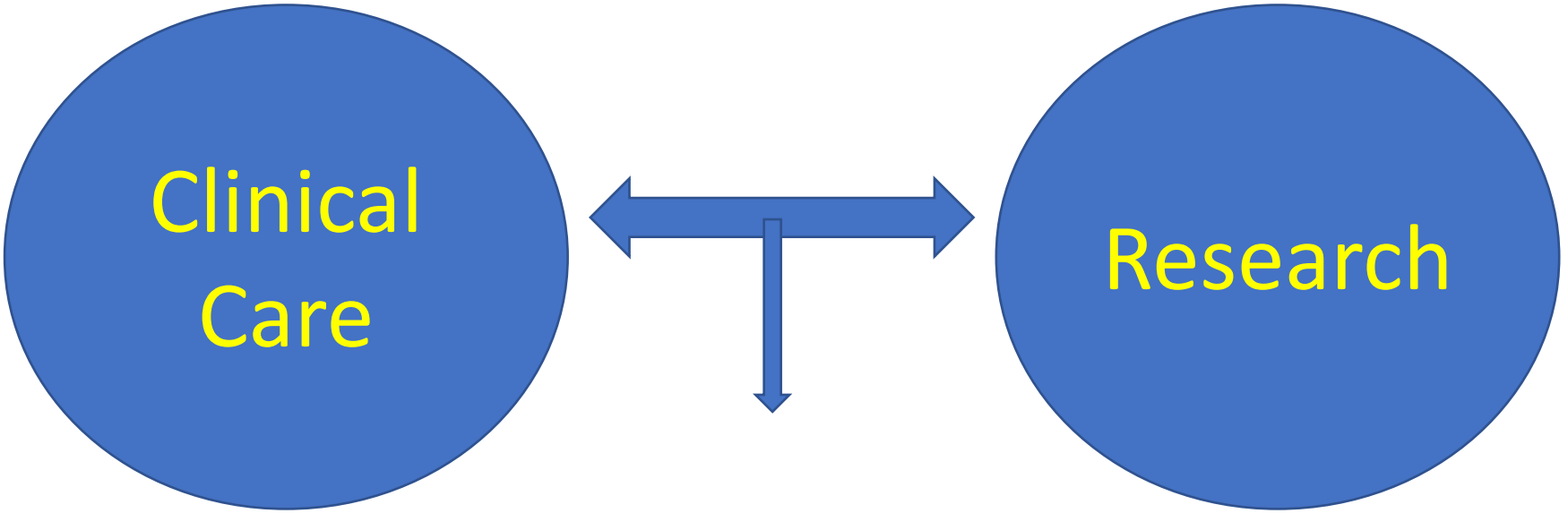
HARVARD
MEDICAL SCHOOL

I have no financial disclosures or conflicts to disclose

Relevant information:

- Member, Board of Directors for the Federation for Children with Special Needs
- Member, Board of Directors for the Down Syndrome Medical Interest Group
- Member, Board of Directors for the National Down Syndrome Congress
- Member, Massachusetts Down Syndrome Congress Scientific Advisory Council
- **My sister, Heather, has Down syndrome**

Clinical Research



What We Know

What We Don't Know

What We NEED To Know
Clinical Research

Agenda

- Pediatric Updates
  - Health Supervision Guidelines
  - Development and Behavior

- Research: Toward Optimizing Neurodevelopmental Outcomes
  - Biomarkers / Investigation of brain patterns in learning
  - JASPER Behavioral Therapy
  - Boston Children's Hospital Longitudinal Database

(A very fast tour)

Part 1: Updates in Pediatric Care

2022 AAP Updated Health Care Guidelines for Children and Adolescents

CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

Health Supervision for Children and Adolescents With Down Syndrome

Marilyn J. Bull, MD, FAAP,[a] Tracy Trotter, MD, FAAP,[a] Stephanie L. Santoro, MD, FAAP,[b] Celanie Christensen, MD, MS, FAAP,[c] Randall W. Grout, MD, MS, FAAP,[d] THE COUNCIL ON GENETICS

2022 Key Updates

- Emphasis on resources, communication
- Prenatal Screening and Diagnosis
  - Prenatal testing – non-invasive cell free DNA testing has most sensitivity / specificity
  - New resources and guidance to improve the ways families receive information
  - Emphasis on non-directed guidance / emotional support
  - Best ways to deliver the diagnosis of Down syndrome
  - Table of guidance for communication with families
- Adolescence guidance and resources: puberty, safety

2022 Key Updates

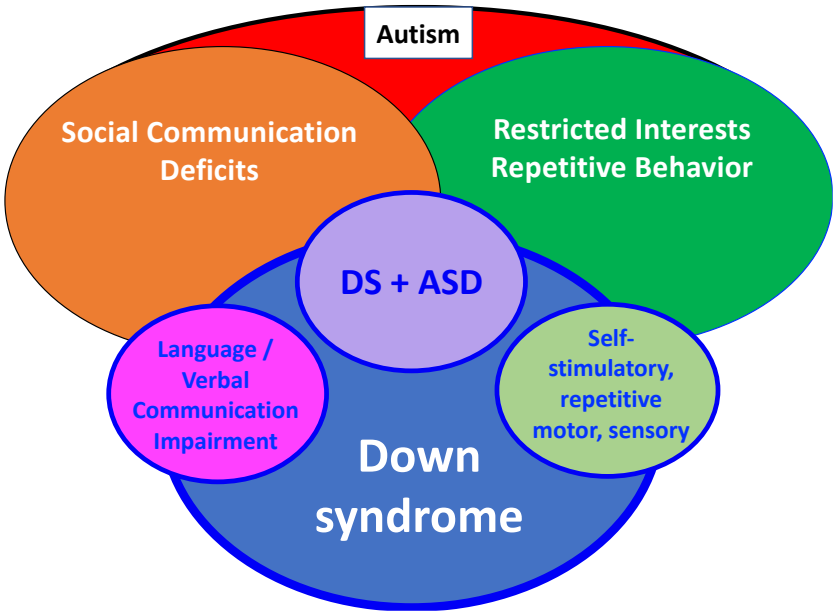
- Minor updates to health recommendations:
  - Sleep study *between* age 3-4 years
  - Screening for Iron Deficiency / Insufficiency
    → Ferritin, TIBC, iron OR ferritin / CRP
  - Monitor for symptoms of leukemia, palpate testes due to increased risk of testicular cancer
  - Increased emphasis on some conditions – feeding difficulty, respiratory infection, dermatologic problems, autoimmune conditions
  - Growth / Nutrition monitoring: New growth curves
    → Use DS-specific BMI growth charts for < 10 years, then CDC charts after 10 years.

2022 Key Updates

- Recognition and emphasis on increased prevalence of co-occurring neurodevelopmental, behavioral, and mental health conditions, and guidance on appropriate diagnosis and treatment
  - Autism Spectrum Disorder
  - Attention-Deficit Hyperactive Disorder
  - Psychiatric
  - Behavior
- Specialized diagnostic evaluations, interventions, support needs

Down Syndrome and Autism

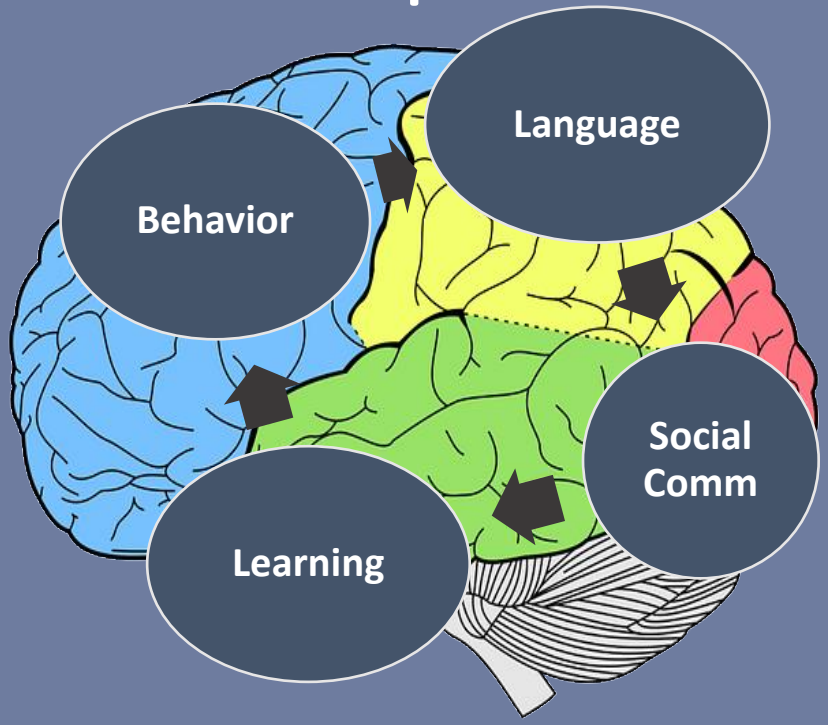
- Prevalence reported 2011 (1%) → 2022 (7-19%)
- Lower cognitive abilities, more difficult with language and communication (verbal and nonverbal), social difficulty, more frequent repetitive behaviors and sensory symptoms, more mood and behavior challenges
- Screen 18-24 months of age, refer to specialized evaluation and interventions



Down Syndrome & Behavioral and Mental Health Needs

- Recognition of phenomenon of unexplained regression in Down syndrome
  - “Acute regression”, “catatonia”, “distintegrative disorder”
  - Need for referral to specialized evaluation
- Developmental and Behavioral Intervention Strategies
- Special considerations for use of psychoactive medications
  - Start Low, Go Slow

Part 2: Research in Learning and Development



Structural and neurophysiological brain differences in Down syndrome may lead to common cognitive and neurodevelopmental characteristics

Research Priorities

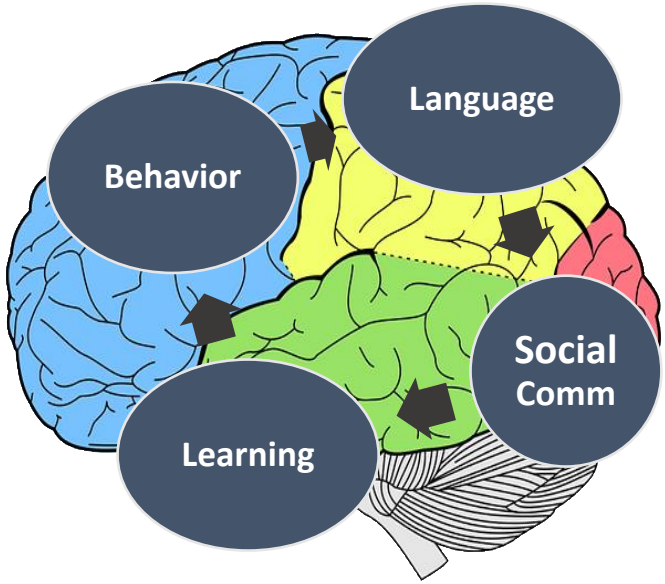
- **What we know:**
  - Common neurodevelopmental characteristics
  - Wide range of abilities: cognition, language, social communication, behavior, functional skills
  - Increased risk of co-occurring neurodevelopmental / psychiatric conditions
- **What we don't know:**
  - What impacts neurodevelopmental outcomes in individuals with Down syndrome? (Early development, behavior, communication)
  - Are there modifiable risk factors?
  - Is there a way to predict neurodevelopment to determine who may need more intensive treatment?
  - What interventions can be used to maximize neurodevelopment and health / wellness?

Research Projects

- What is happening very early on in brain development?
- Is there a way to predict neurodevelopmental outcomes in babies with Down syndrome?
- Can we intervene to positively impact the developmental trajectory In children with Down syndrome?

Early Detection, Understand Mechanisms → **Targeted Treatment** → **Improved Outcomes**

Can EEG serve as a brain-based risk marker and help us understand mechanisms of development, and predict who may have higher risk?



NIDCD (NIH INCLUDE): Down Syndrome Infant Screening Project
Tapley Family Fund: JASPER Down Syndrome

Joint Attention Symbolic Play Engagement Regulation

Teach	Teach skills through modeling and imitating
Increase and Improve	Increase and improve skills that impact engagement • e.g. coordinating attention, flexibility in play skills, achieve higher play levels
Provide	Provide opportunities for learning and social communication through play
Increase	Increase emotional and behavioral regulation
Decrease	Decrease self-stimulatory behaviors

Clinical Trial of Behavioral Therapy in Down Syndrome

- JASPER: parent mediated behavioral therapy intervention for children targeting communication, social skills, play, behavior
- Neurobehavioral Assessments: Development, Language, Socialization, Behavior
- Caregiver strategies, self-efficacy, hassles/stress
- Neurophysiological Assessments: EEG, ERP

Research → Evidence → Advocacy

Known strategies and therapies may *already* exist that individuals with Down syndrome do not have consistent access to b/c not rigorously studied in Down syndrome

We hope that by demonstrating effectiveness through rigorously designed clinical trials that we can change the landscape of available interventions for individuals with Down syndrome

Longitudinal Database

- Boston Children's Hospital developed a systematic, standardized way to gather clinical information from parents / families (n=900 unique patients)
  - Standardized documentation of medical conditions, services / interventions, and development / behavior of clinical information
  - Longitudinal data collected every 6 months during clinical visits
- Goals:
  - Exploration of patterns of development
  - Investigate impact of medical conditions on development and learning
  - Learn about what interventions are effective and help maximize neurodevelopment, health, and wellness

Supported by Tapley Family Fund

- **Mission:** To partner with families, interdisciplinary providers, and researchers to evaluate, treat and investigate medical, cognitive, neurodevelopmental, educational, mental health, behavioral and social needs of individuals with Down syndrome from the time of diagnosis until transition to adult care.
- **Vision:** All individuals with Down syndrome are physically and emotionally healthy, engaged at their highest capacity at school or vocation, have meaningful family and social relationships, and are valued members of their communities.