

# 2025 DSMIG-USA Annual Symposium

## Novel Screening Tool to Identify Down Syndrome-associated Arthritis in Clinical Practice

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## Disclosures

- Consulting for Pfizer Pharmaceuticals

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# Introduction

- Children with DS have **increased frequency**:
  - Infection
  - Hematologic malignancies
  - Autoimmune disease
  - Inflammatory arthritis
    - Down syndrome-associated arthritis (DA)

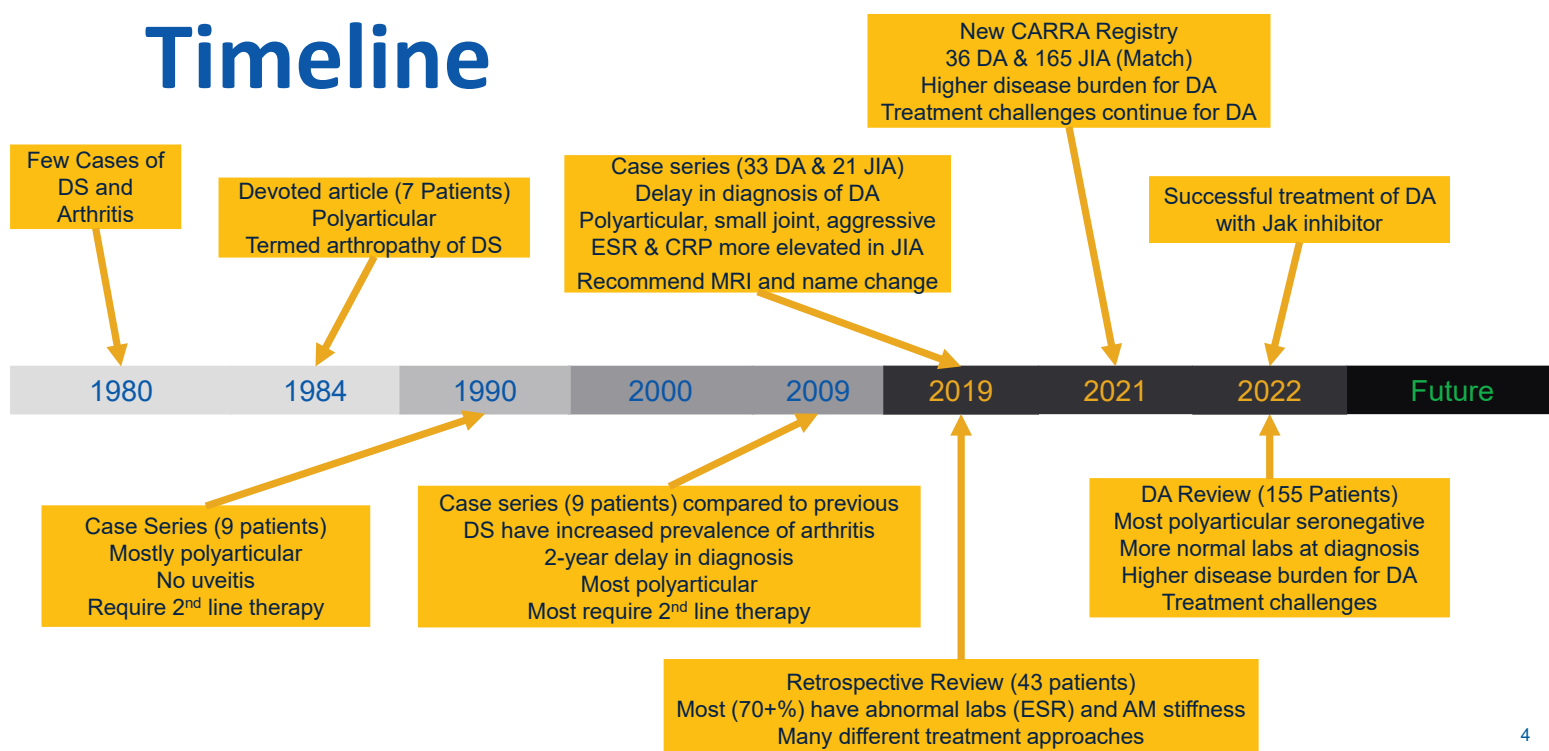
## Inflammatory diseases

Coeliac disease	Increased
Thyroid disease	Increased
Type I diabetes mellitus	Increased
Alopecia areata	Increased
Arthritis (rheumatoid arthritis, juvenile idiopathic arthritis, Down's arthropathy)	Increased
Psoriasis	Unchanged
IBD (Crohn's disease, ulcerative colitis)	Unchanged
Hidradenitis suppurativa	Increased
Kawasaki disease	Decreased
Osteoarthritis	Increased
Atherosclerosis	Decreased
Alzheimer's-like dementia	Increased

Verstegen RHJ, et al. *Pediatr Allergy Immunol.* 2019



## Timeline



# Health Supervision for Children and Adolescents With Down Syndrome

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

	Prenatal	Birth–1 mo	1 mo–1 y	1–5 y	5–13 y	13–21 y
Counseling regarding prenatal screening test & imaging results						
Plan for delivery						
Referral to geneticist						
Parent-to-parent contact, support groups, current books and pamphlets						
Physical exam for evidence of trisomy 21						
Chromosomal analysis to confirm dx						
Discuss risk of recurrence of Down syndrome						
Echocardiogram						
Radiographic swallowing assessment if marked hypotonia, slow feeding, choking with feeds, recurrent or persistent respiratory sx, FTT						
Eye exam for cataracts						
Newborn hearing screen and follow-up						
Hx and PE assessment for duodenal or anorectal atresia						
Reassure parents delayed and irregular dental eruption, hypodontia are common						
If constipation, evaluate for limited diet or fluids, hypotonia, hypothyroidism, GI malformation, Hirschsprung			Any visit			
CBC to R/O transient myeloproliferative disorder, polycythemia						
Hb annually; CRP & ferritin or ChR if possible risk iron deficiency or Hb <11 g				Annually		
Hemoglobin						Annually
TSH (may be part of newborn screening)			6 and 12 mo	Annually		

- 2011 – No mention of arthritis or screening for arthritis.
- 2022 – JIA mentioned once (prevalence)

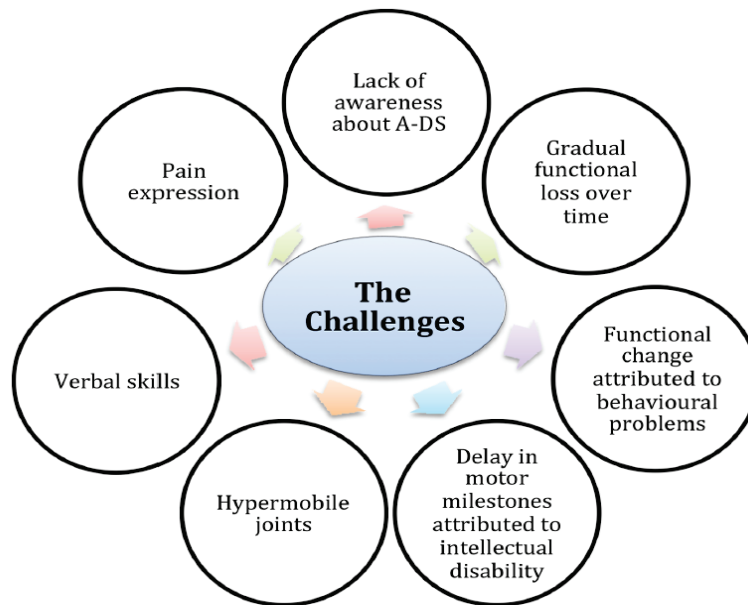
#### Autoimmune conditions

Hashimoto thyroiditis	13–39
Graves' disease	1
Celiac disease	1–5
Type 1 diabetes	1
Juvenile idiopathic arthritis	<1
Alopecia	5

## Screening for Down syndrome-associate Arthritis

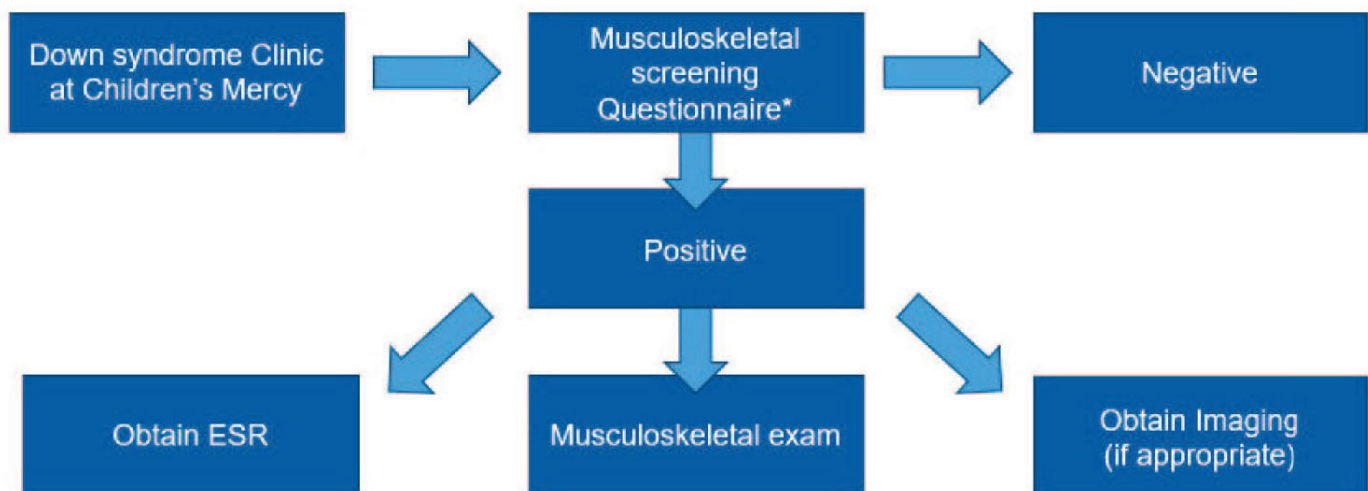


# Challenges with Screening



Foley CM, et al. *RMD Open*. 2019; 5(1):e000890

## Down syndrome Clinic screening



\* Two questions:

1. In the past 3 months, have you noticed any joint swelling?
2. In the past 3 months, have you noticed any morning stiffness?

## Down syndrome Clinic screening

- 91 total patients with Down syndrome (DS) screened
- 4 positive
  - 1 confirmed arthritis (positive US findings), normal ESR
  - 1 questionable arthritis (normal XR), normal ESR
  - 1 no arthritis (normal XR), normal ESR
  - 1 no arthritis (normal XR), normal ESR – abnormal exam due to stroke
- 1 true positive out of 91 total (1%)

**14% (13/91) of parents were aware of risk of arthritis in DS**

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Jones JT, et al. *Glob Pediatr Health*. 2021; 7(8):2333

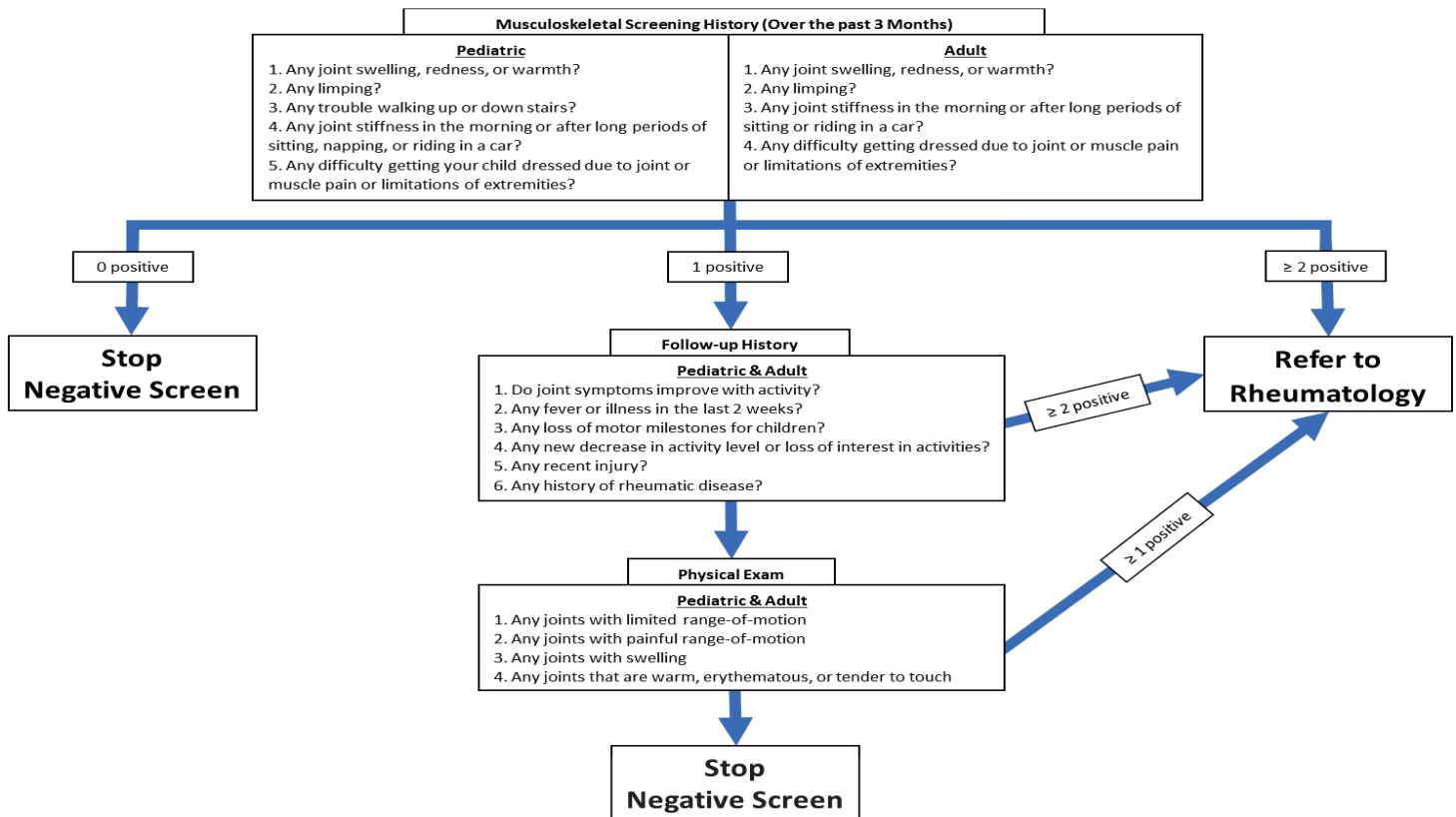


## Down Syndrome Screening Tool

- DS-musculoskeletal screening tool - developed and piloted
- Multi-center, international study across 8 institutions
- All sites were trained on MSK exam
  - Normal joint range-of-motion in DS
- Feasibility assessed

Jones JT, et al. *Am J Med Genet C Semin Med Genet*. 2023 Dec; 193 (4): e32076

## Down Syndrome Musculoskeletal Screening Tool



# Down Syndrome Screening Tool

Table 2. Positive Screen and Diagnosis Rate\*\*\* of Down syndrome-associate Arthritis

Site	n	Positive Screen Rate n (%)	Diagnosis Rate n (%)
University of Queensland	12	3 (25.0%)	1 (33.3%)
Pediatrico Bambino Gesù	427	16 (3.8%)	2 (12.5%)
Boston Children's Hospital*	306	7 (2.3%)	1 (14.2%)
Children's Hospital Los Angeles	107	24 (22.4%)	16 (66.7%)
Children's Mercy Kansas City	76	2 (2.6%)	1 (50.0%)
Duke University Medical Center	8	1 (12.5%)	0 (0%)
Massachusetts General Hospital**	115	9 (7.8%)	0 (0.0%)
Children's Hospital of Pittsburgh	60	0 (0.0%)	0 (0.0%)
<b>Total</b>	<b>1111</b>	<b>62 (5.6%)</b>	<b>21 (33.9%)</b>

\* 1 patient screened positive and has a missing diagnosis

\*\* 5 patients screened positive and have a missing diagnosis

\*\*\* Positive Predictive Value



# Down Syndrome Screening Tool Results

- 1111 patients evaluate, average age 10 yrs (SD 6.7)
- 6% positive screens and 34% diagnosed with DA.
- Joint swelling, redness, warmth, joint stiffness, and painful range-of-motion were most associated with a diagnosis of DA.
- Feasibility – 5 minutes for history, 10 minutes for exam



## Takeaway Points

- Down Syndrome-specific MSK screening tool successfully identified new cases of DA
- Feasibility was acceptable
- 2% prevalence for DA

# Questions?

## Acknowledgments

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### Collaborators

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