DS Participant Meeting

Beau Ances, MD, PhD
Daniel J Brennan MD Professor of Neurology
Saturday, February 26, 2022
Thank you for joining us today for our First Annual Participant Meeting!

This presentation is being recorded, if you don’t want your video and/or name to appear in the recording, please please turn off your video.

Please remain muted throughout the presentation, you can unmute for the question/comment session at the end.

You can type your questions in the chat, please mention the name or organization if you have a specific question.

If we don’t get to your question, please email Brittany at bmnelson@wustl.edu and I will work to find your answers.

If you registered for the zoom and you selected to be contacted in the future, we will be contacting you with study related information. You can always opt out by emailing Brittany at bmnelson@wustl.edu
Today’s Outline

- Review of study data from previous DS research
  - Beau Ances, MD, PhD

- Introduction into new available DS research studies
  - Beau Ances, MD, PhD

- Introduction of community programs:
  - Washington University DS Clinic
    - Liz Hall, NP
  - Down Syndrome Association of Greater St. Louis (DSAGSL)
    - Erin Suelmann, MA, MPH and Makini Anwisye
  - St Louis Arc (SLARC)
    - Mary Anne Tolliver and Sharon Spurlock
  - Pujols Wellness Center
    - Stacey Laughlin, LMSW
  - Artists First
    - Sheila Suderwalla

- Comments and Questions (please use chat feature for questions and we will get to them at the end of the talk)
Study Results
Amyloid PET in Adults with DS

There exists considerable variability regarding conversion
Tau PET in Adults with DS

Tau PET is typically elevated in DS adults who are amyloid PET positive

Rafii et al., 2017; Zammit et al., 2020
Comparison of Neuroimaging Signatures of AD in Adults With DS

Fortea et al., 2020
Head and Ances, 2020
- 5 drugs have been approved for late onset AD (Tacrine, Donepezil, Galantamine, Exelon, Memantine)
- Studies for donepezil in adults with DS are mixed. Most studies have had a relatively small sample size and shown modest to no effect with some demonstrating adverse side effects.
- A more recent study of memantine in adults with DS showed no improvement but also no increase in adverse side effects.
- Galantamine – no studies have been performed to date
- Tacrine – no studies have been performed to date
Proposed Cascade of Biomarkers for ADAD and DSAD

ADAD

DSAD

Bateman et al., 2011

Fortea et al., 2020
Comparison of Amyloid PET With CSF Aβ42/40 Ratio in ADAD and DSAD

DIAN mutation-carriers (MC)

ABC-DS

Boerwinkle et al., to be submitted
Amyloid PET As A Function of Cognitive Status in ADAD and DSAD

Boerwinkle et al., to be submitted

aDS = asymptomatic DS
sDS = symptomatic DS
aMC = asymptomatic MC
sMC = symptomatic MC
Amyloid PET as function of age for ADAD and DSAD
Regional Changes in PET Amyloid as a Function of EYO of ADAD and DS
This video shows the accumulation of amyloid in different regions of the brain over time.

- The top row shows this in individuals with autosomal-dominant Alzheimer's disease.
- The middle row shows individuals with Down syndrome.
- The bottom row is the difference between the two.

Boerwinkle et al., *to be submitted*
New Research Opportunities

ABC-DS
Lifestyles
TRC-DS
DS Umbrella
This study aims to identify biomarkers for Alzheimer disease in adults with DS. We hope to identify individuals who are at increased risk for developing the disease later in life.

We believe changes in the brain occur 10-15 years before an individual or caregiver notices changes in behaviors or memory.

Alzheimer disease is due to the buildup of proteins which are building blocks of our body. In particular, amyloid and tau deposit in the brain. Over time, increases in these proteins lead to changes in how the brain works and communicates.

We believe adults with Down syndrome are at a greater risk for developing Alzheimer disease because the 21st chromosome has a gene that codes for the amyloid protein.

Because individuals with Down syndrome have an extra 21st chromosome, they have more amyloid deposited in the brain compared to someone without DS.
ABC-DS is a multidisciplinary, multi-site longitudinal study that examines biomarkers of AD in a large cohort of adults with DS.

- It is comprised of 8 Locations across the United States and one in the United Kingdom
- The ABC-DS study sites plan to recruit and follow a total of 550 individuals and 50 sibling controls.
- ABC-DS is funded by NIH and includes multiple different institutes: NIA, NICHD, and the INCLUDE Project
Study Details

- We are actively recruiting adults with Down syndrome who are ages 25 years and older.
- Assessments are every 16 months over 5 years.
- Study visits take place at Washington University in St. Louis.
- Participants are compensated for their time (each procedure completed).
- Reimbursements are provided for mileage, parking, and stipends for meals and lodging (if traveling from a distance).
- Our plan is to enroll 45 adults with Down syndrome and 10 sibling controls.
Study Procedures

- Consent (assent) process
- Medical History
- Physical/Neurological Examination
- Cognitive testing with Participant
- Interviews with Study Partner (Caregiver)
- Blood Draws – examining genetics, fats and proteins in blood
- MRI Scan – uses a magnet to help us look at brain structures non-invasively
- PET Scans – inject a small amount of radiotracer that allows us to look at the amyloid and tau proteins in the brain
- Optional: Lumbar Puncture to look at spinal fluid that surrounds the brain and spinal cord
## Schedule of Events

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<th>Month 0</th>
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Study Objective: Development of a well-characterized, trial-ready cohort to facilitate rapid enrollment into AD treatment and prevention trials for adults with DS.

By the age of 35 years old, many adults with DS will already have amyloid plaques – a hallmark of AD.

Preliminary data show that adults with DS exhibit significant changes in AD-related biomarkers between the ages of 35-55. It is also within this age range that AD-related cognitive decline and dementia typically manifest in DS.

AD is the primary cause of death in adults with DS over the age of 35. A need exists for including adults with DS in clinical trials for AD.
Enroll 120 non-demented adults with DS across 15 sites into the TRC-DS.

Washington University in Saint Louis is one of 7 sites in the US participating in both ABC-DS and this study.

We will be enrolling ~ 10 DS participants.

- Bloomington, IN
- Boston, MA*
- Boulder, CO
- Cleveland, OH
- Irvine, CA*
- Lexington, KY*
- Madison, WI*
- Nashville, TN
- Phoenix, AZ
- Pittsburgh, PA*
- Staten Island, NY*
- St. Louis, MO*

*Indicates co-enrolling site for ABC-DS and TRC-DS
Study Details

- Recruiting individuals with Down syndrome ages 35 to 55 years old.
- Assessments will be every 16 months.
- Study visits take place at Washington University in St. Louis.
- Participants are compensated for their time (each procedure completed).
- Reimbursement will be provided for mileage, parking, stipends for meals and lodging (if traveling from a distance).
Study Procedures

- Consent (assent) process
- Medical History
- Physical/Neurological Examination
- Cognitive testing with Participant
- Interviews with Study Partner (Caregiver)
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This study examines the effect of four lifestyle factors – physical activity, sleep, cognitive stimulation, and social engagement – on development of AD and cognitive decline in adults with DS.

Growing evidence from non-DS populations implicates poor physical activity, sleep, cognitive stimulation, and social engagement can contribute to the early development of AD.

The majority of studies on lifestyle factors, however, have focused on the later clinical stages of AD.

Thus, across DS and non-DS populations, there is a critical need for studies to examine the effects of lifestyle factors on developing AD.
This is a supplemental study for ABC-DS and participants must be co-enrolled or plan to be enrolled in ABC-DS.

A total of 140 adults with Down syndrome will participate.

A total of 3 sites are participating:
- University of Wisconsin-Madison
- University of Pittsburgh
- Washington University in St. Louis.
Daytime physical activity will be assessed continuously using an Actigraph that is worn for 7 days continuously.

A Sleep Diary will be completed daily. The caregiver will help complete the forms with the input from the DS participant.

On the first night (or an alternative night if preferred by the participant), the adult with Down syndrome will also wear the WatchPAT 300 sensors:

- This device uses a peripheral arterial tone signal to assess for sleep disordered breathing, and thus does not wear anything to be worn over the face.
- This device will only be worn for ONE night.
- Results of sleep study will be available for participant and their family.
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DS Umbrella Study

- Designed for individuals who are unable to complete the tasks involved in ABC-DS or TRC-DS and for individuals who are interested in completing other procedures.
- Limited cognitive assessments
- Annual Visits
- Funded by Beau Ances, MD, PhD Discretionary Fund
Study Procedures

- Consent (assent) process
- Medical History
- Physical/Neurological Examination
- Limited Cognitive testing with Participant
- Interviews with Study Partner (Caregiver)
- Blood Draws
- Stool Samples
## DS Umbrella Study

### Components by Visit

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Community Organizations
What’s available for individuals with Down syndrome and their families
Down Syndrome Clinic:
Providing Comprehensive Care for Patient and Family

Elizabeth Hall, NP
Washington University School of Medicine
St. Louis Children’s Hospital
Down Syndrome Center
St. Louis Children’s Hospital

- Established in 2005
- Multidisciplinary clinic
- Currently >500 visits per year
- Birth through adulthood
- 4 in-person half days per week, plus telemedicine
  - St Louis Children’s Hospital
  - Children’s Specialty Care Center West County
  - Children’s Specialty Care Center South County
- Adult Down syndrome Clinic established in 2021
Mission

• Maximize health by understanding and anticipating known health issues and risks.
• We strive to optimize the quality of life for people with DS and their families by offering comprehensive, individualized care in one setting
Major Health Concerns affecting individuals with DS

- Cardiology
- Gastroenterology
- Ophthalmology
- Audiology and ENT
- Hematology
- Immunology
- Orthopedics
- Neurology
- Urology
- Endocrinology
- Dermatology
- Pulmonary/Sleep Issues
Developmental and Behavioral Differences

- Delays in certain areas of development
  - Gross and Fine motor
  - Cognitive
  - Language
    - Expressive delays > Receptive
- Excel in some areas of development
  - Social
  - Visual memory
- ADHD
  - Management issues
- Autism
- Sensory Integration Disorders
- Regression
Contact Information

• Medical Director: Dr. Kathy Grange
• Clinic Nurse Coordinator: Dawn Easterlin 314-273-7992
• Nurse Practitioners: Liz Hall 314-454-4832 and Laura White
• Genetic Counselor: Rachel Slaugh
• Genetics Main 314-454-6093
Who is the Down Syndrome Association of Greater St. Louis?
• Established in 1976 by a small group of parents
• Celebrated our 45th birthday on June 7th 2021
• Serve 1900 families in the greater St. Louis region (approximately 100-150 mile radius)
• Headquartered in Brentwood
• 11 staff – 4 PT & 7 FT
Mission:
DSAGSL serves, supports, and celebrates the lives of individuals with Down syndrome and their families, through every stage of life.

Vision:
An inclusive community in which all people with Down syndrome can reach their full potential.
SERVICES OFFERED BY DSAGSL:

**Family Support & Workshops**
Programs and services for parents, caregivers, and family members of people with Down syndrome
 Read More »

**Small Steps**
Programs and services for individuals with Down syndrome ages 2-8
Read More »

**Next Steps**
Programs and services for individuals with Down syndrome ages 8-21
Read More »
SERVICES OFFERED BY DSAGSL

Ready 2 Work Employment
Programs and services for individuals with Down syndrome ages 21+ including individuals looking for employment
Read More »

Silver Steps
Programs and services for aging individuals with Down syndrome and their caregivers
Read More »

Advocacy & Awareness
The DSAGSL is involved in raising public awareness as well as advocating for the rights, dignity, and respect of individuals with Down syndrome in a variety of ways including hosting events, speaking with local news, and contributing to advocacy groups in the St. Louis area.
Read More »
SERVICES OFFERED BY DSAGSL

21 Club & Social Events
This social group is for individuals of all abilities 21+. Hosted by the Young Friends Board of DSAGSL, the social activities are an opportunity for individuals with Down syndrome to socialize with typical peers, fostering inclusion, acceptance, and understanding.

Read More »

In-Person & Virtual Programming
Virtual Programs through Zoom, Facebook, and YouTube. A variety of programs are available for all ages and abilities.

Read More »

Summer 2021 Programs
Summer Programs 2021 We’re finally getting back together in person this summer! Check out what we have planned for all ages! Please contact Andi McCormick

Read More »
SERVICES OFFERED BY DSAGSL

DSAGSL Virtual & In Person Programming
JANUARY - MARCH 2022

MON
9:00AM Music & Movement with Andi
Facebook

TUE
12:00PM Let's Talk!
Meet up for teens and adults
Zoom

WED
1:00PM Living Your BEST Life
Facebook

1:00PM Stand Up, Sit Down
with Corny the Clown
Facebook

THU
10:00AM Improv!
Zoom
Register: don@dsagsl.org

12:30PM Express Yourself!
Group speech therapy for adults
Zoom

10:00AM Fusion Fridays!
Group Activities
Facebook
February 10a-12p
ZOOM ONLY!

FRI
10:00AM In Person
starting: TBD
Facebook
Register: don@dsagsl.org

Find ALL our Virtual & In-Person Programs at www.dsagsl.org/programming
SERVICES OFFERED BY DSAGSL
CONNECT WITH DSAGSL

Facebook.com/DSAGSL  Instagram.com/DSAGSL  DSAGSL.org
For more information

Down Syndrome Association of Greater St. Louis
1300 Strassner Drive | Brentwood, MO 63111
314.961.2504 | info@dsagsl.org | www.dsagsl.org
Services

FAMILY SUPPORTS

INDIVIDUAL SUPPORTS
Family Support

Family Workshops
Video Learning Library
Information & Referral
Prevention

Parent Cafe
Support Groups
SibShops
Advocate Groups

Family Navigation
Respite
Individual Services
Children's Services

Community
Embedded Services

Capable Kids
& Families

Parents Learning
Together
Transition to Adulthood

What will I do as an adult?

Further Education

Employment/meaningful day

Community living skills
Adult
Community Engagement
LIFT
Supported Employment
Leisure Services
Behavior Supports
Residential
AGING
Residential Services
Aging With Empowerment
Nutrition Center
Albert Pujols Wellness Center for Adults with Down Syndrome
The Albert Pujols Wellness Center for Adults with Down Syndrome was the first of its kind in the area that specifically addressed the need for wellness services and programs dedicated solely for adults with Down syndrome. The Center opened its doors in November 2009.
Services and programs provided at the Wellness Center
The Wellness Center’s Clinic

Medical Director:
Dr. Cynthia Poelker
Our Center provides serves to individuals 17 years of age and older

- Comprehensive medical examination with a focus on:
  ~ Transitional evaluations from pediatric care
  ~ New patient evaluations

- Referrals to specialists as needed

- Psychosocial assessments by a licensed social worker

- Nutritional Assessments with a registered dietician
The other part of our Wellness Center: Our classes and programs

For all new patients, a wellness visit must first take place prior to signing up for our classes
Some of the programs we have offered at the Wellness Center

- Socialization programs (Friendship class)
- Fitness programs (Yoga, Kickboxing, strength training, cardio, balance training)
- Tai Chi
- Country Line Dancing
- Speech/Language programs
- Nutrition programs (cooking classes, portion control, Eat this not That, Fitness Focused)
- Music programs (Glee choir, hand chime choir, musical instrument groups)
- Arts and crafts programs
- Book Club
Growth Over Time
COVID: A Challenging Time

- In-person classes had to be put on hold
- So, how do we keep our clients engaged
- Like so many other agencies you had to think outside of the box
Online classes (virtual) has it’s own unique problems

- Connectivity issues
- Hearing & Vision issues
- Some clients can’t stay engaged with virtual programs
Why we do what we do
Janice Scully: parent in her own words

There are so few opportunities for our children with Down Syndrome to develop meaningful relationships with their peers. This center provides friendships and the sense of belonging to a community. Whether an individual has the verbal skills to carry on a conversation is unimportant. Just a smile and “hello” from their friends is enough to bring a smile to their faces.

The classes allow an exploration of skills that are rarely available to many adults with disabilities. The support of the staff is wonderful. Our children can learn and grow at their own abilities with guidance, fun and kindness.

For the parents, this center is an anchor. It is our therapy sessions and proof that we are not alone. Someone will always have a helpful comment or suggestion on how to solve a problem. We gladly share funny stories, challenges and pride for our children.

The St. Luke’s community have given us the opportunity to develop lifelong friendships with the clients and parents while encouraging healthy adults with Downs Syndrome.
Why we do what we do (cont.)
Paige and Shari Brune: client and parent in their own words

First we would like to thank the Pujols Wellness Center for Adults with Down Syndrome and the hard work and dedication of Stacey and the staff at the center as well as all of the volunteers that make this operation run smoothly.

Paige has been involved with the Center for 11 years and we have seen the program grow and develop as the needs of the community have also developed. From a parent perspective, this Center has become a continuation of Paige’s education and development post Highschool and vocational training.

The Center not only offers Paige a time of social engagement (even with COVID through WebEx classes) but, a chance to learn new skills. From Music & Art, Speech & Language, Health & Wellness and awesome Exercise classes, the Center has introduced many areas of extended learning and socialization.

Paige has so enjoyed expanding her love of music and art. The Hand chimes and Glee classes are always a highlight of her week. Learning to enjoy playing music and singing with others always brings a sense of joy and confidence to her life!! Paige also looks forward to the Craft Classes! No matter what project the group is working on Paige really gets into it and has fun. Stacey has been so creative in getting these projects together keeping in mind the skills needed to complete the project.

Paige has also enjoyed volunteering at the Center on occasion with mailing out flyers to helping organize the craft closet, she really enjoys helping others.

For many parents, the Center also serves as a meeting place to share ideas, communicate needs and be a true support for each other. It’s such a wonderful feeling knowing that you can talk to parents that have either been through the same challenges or are facing the same challenges as you are. The Center has offered seminars on Estate Planning, Health in the Down Syndrome Community and others that have also enriched our lives which in turn enriches the adults we have in our care.

We feel so Blessed that the Center is available and has become such an important part of our lives as well as the lives of many Adults with Down Syndrome. When we talk to other families that do not live in this area, we are so proud to talk about the Wellness Center and what it means to us through programs and a true sense of community for our family. Again, we appreciate all that the Center does for us and Paige.

Thank you for all you do!
Kendall, Shari & Paige Brune
THANK YOU!
Artists First is an inclusive, non-profit art studio empowering artists of all abilities.
The Integrated Adult Open Studio provides participants with a studio stocked with professional supplies and technical facilitation by professional artists.

Youth with and without disabilities in the Integrated Youth Open Studio collaborate in a diverse environment where they can develop artistic and social skills.

Artists First for Veterans is a nontraditional creative support program for current and former Armed Services members that supports the development of positive self-identity, self-expression and community re-integration.
OMA is an evidenced based intergenerational visual art program for people with memory loss.

Lifeworks is a program tailored to professionally minded artists with developmental disabilities to obtain their greatest potential and growth in employment, social, emotional and physical domains through the arts.

Artists First's In View Mural Project provides the community (businesses, residences, and public spaces) with high quality, hand-painted, custom murals. The project provides artists with disabilities, veterans, and at-risk youth with confidence-building opportunities, work experience, inclusion, and visibility.
CREATIVE SELF-EXPRESSION:

Contributes to the improvement of mental health in Individuals with developmental disabilities (Grbić et al., 2018).

Improves cognitive, sensory-motor functions, fosters self-esteem, self-awareness, cultivates emotional resilience, promotes insight, enhances social skills, reduces and resolve conflicts and distress, and advance societal and ecological change (American Art Therapy Association, AATA, June 2017).

Participatory art making offers the “means by which [people] acquire the skills, language and connectedness” (National Organization for Arts in Health. 2017).
ARTISTS FIRST
St.

Art For All

7190 Manchester Road
Maplewood, MO 63143

info@artistsfirststl.org

www.artistsfirststl.org
ABL is Committed to the Saint Louis DS Community
We would like to extend a huge thank you to the organizations who have joined us today:

- WUSM Down Syndrome Clinic
- Down Syndrome Association of Greater St. Louis
- St Louis Arc
- Albert Pujols Wellness Center for Adults with Down Syndrome
- Artists First

To all the past and future participants or clinic patients and families, we’re so happy you could join today and look forward to working with you in the future. We can’t do this without you!
Questions and Answers

- You can type your questions in the chat, please mention the name or organization if you have a specific question.
- If we don’t get to your question, please email Brittany at bmnelson@wustl.edu and I will work to find your answers.
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