

Moving forward: An overview and review of TRIS project activities (July 2021-June 2025)



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SOFT Conference
July 2025



Began in 2007, the Tracking Rare Incidence Syndromes (TRIS) project seeks to increase awareness and knowledge for families and professionals touched by rare trisomy conditions and aims to facilitate improved decision making for optimal services and supports for affected children and their families.

TRIS project components

- TRIS surveys
 - Full survey (baseline survey) collects information on birth history, medical issues, developmental progress and support needs
 - Modified survey (up to 60 days)
 - Follow-up Survey (annual updates on key items from the TRIS baseline survey)
- Responding to requests for medical and/or developmental information
- Outreach (raising awareness, recruitment)
- Dissemination of survey results

In review

- Survey completions
- Awareness and recruitment activities
- Publications and presentations
- Collaboration activities
- Requests from parents, students and professionals (medical, teachers etc.)
- Ongoing activities

Survey completions

Completions as of July 1, 2025:

Full TRIS Survey = 612 (n=1075, 57%)

Modified TRIS Survey = 252 (n=282, 89%)

TRIS Follow-up Survey completions

Year 1 = 424

Year 2 = 343

Year 3 = 287

Year 4 = 228

Year 5 = 174

Year 6 = 136

Year 7 = 105

Year 8 = 80

Year 9 = 60

Year 10 = 39

Year 11 = 33

Year 12 = 26

Year 13 = 16

Year 14 = 10

Year 15 = 6

Year 16 = 3

Awareness and recruitment activities

- Respond to posts and call outs on trisomy related Facebook pages including SOFT, Rare Trisomy Parents, Trisomy 9 Mosaic Families, and Facebook Messenger
- Responding to emails requesting information
Google Alerts for “Trisomy in the News”
- Parent to parent contact

Encourage all interested to enroll in the project
<https://tris.siu.edu/survey/form/PreEnroll.php>

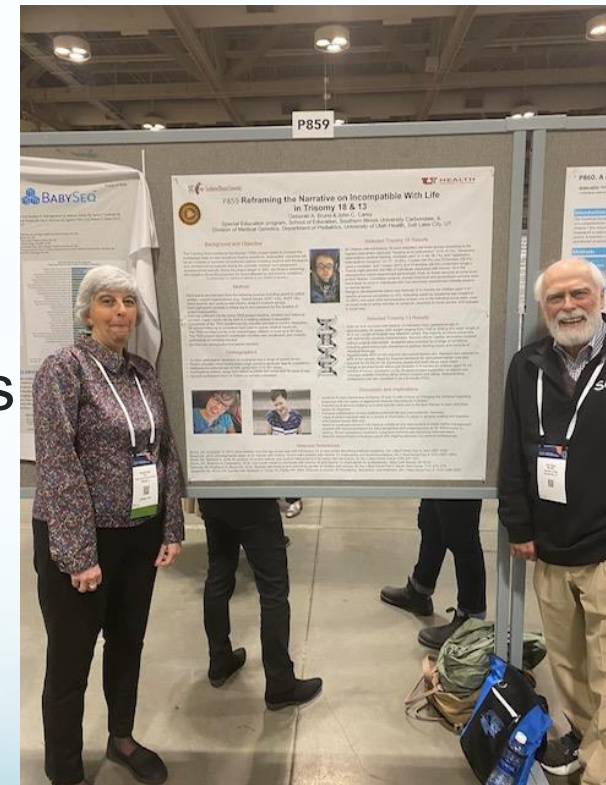
TRIAD in trisomy 8 mosaicism

Continuing work with a researcher at the National Institutes of Health

- Manthiram K, Bowes M, Wu Z, Shi L, Xu Q, Gao G, Abdel Elkahlon A, Oguz C, Kalsi S, Dulau-Florea A, Cassini T, Pak E, Dutra A, Deutch N, Perazzio S, Romeo T, **Bruns D**, ...& Kastner D. (2023, May). *Trisomy 8 associated autoinflammatory disease is characterized by dysregulated myeloid cells*. Annual Meeting of the International Society of Systemic Auto-inflammatory Diseases, Toronto, Canada. [Assisted to prepare, did not attend to present]
- Manthiram K, Bowes M, Wu Z, Shi L, Xu Q, Gao G, Abdel Elkahlon A, Oguz C, Kalsi S, Dulau-Florea A, Cassini T, Pak E, Dutra A, Deutch N, Perazzio S, Romeo T, **Bruns D**, ...& Kastner D. (2023, May). *Trisomy 8 associated autoinflammatory disease is characterized by dysregulated myeloid cells*. Immunology2023 Annual Meeting, Washington, DC. [Assisted to prepare, did not attend to present]
- Manthiram, K., Xu, Q., Wu, Z., Bowes, M., Gao, S., Oguz, C., ... **Bruns, D.** ...& Kastner, D. (2023). Trisomy 8-associated Autoinflammatory Disease (TRIAD) is characterized by dysregulated myeloid cells. *The Journal of Immunology*, 210(1 Supplement), 155-07.

ACMG (March 2024)

Bruns, D. A., & Carey, J. C. (2024, March). *Reframing the Narrative on Incompatible With Life in Trisomy 18 & 13* (Top Ten Ranked). Poster session for the American College of Medical Genetics Annual Clinical Genetics Meeting, Toronto, Canada.



Seizures in t18

- Jaspersen, S. L., **Bruns, D. A.**, Candee, M. S., Battaglia, A., Carey, J.C., & Fishler, K. P. (2023, March). *Seizures in trisomy 18: prevalence, description, and treatment*. Poster for American College of Medical Genetics, Salt Lake City, UT. [Assisted to prepare, did not attend to present]
- Jaspersen, S. L., **Bruns, D. A.**, Candee, M. S., Battaglia, A., Carey, J. C., & Fishler, K. P. (2023). Seizures in trisomy 18: Prevalence, description, and treatment. *American Journal of Medical Genetics, Part A*, 191(4), 1026-1037.

Invitation from Tohoku

Kosho (Department of Medical Genetics, Shinshu University Hospital)

Bruns, D. A. (2024, November). *An overview of the TRIS project: Focus on parent generated data.* Invited session for Trisomy 18 Support Group. Matsumoto, Nagano Prefecture, Japan.

Bruns, D. A. (2024, November). *Changing the narrative on trisomy 18 and 13 through an international database.* Invited session for 68th Meeting of the Japanese Society of Neonatal Development and Medical Sciences, Matsumoto, Nagano Prefecture, Japan.



Trisomy 9 mosaicism

Largest sample described in the literature (n=101):

Bruns, D. A. (2025). Presenting characteristics and medical conditions in 67 cases with trisomy 9 mosaicism. *Clinical Genetics*.
<https://doi.org/10.1111/cge.14742>

Bruns, D. A. & Campbell, E. (2015). Twenty-five additional cases of trisomy 9 mosaic: Birth information, medical conditions, and developmental status. *American Journal of Medical Genetics Part A*, 167(5), 997-1007. DOI: 10.1002/ajmg.a.36977

Bruns, D. (2011). Presenting physical characteristics, medical conditions and developmental status of long-term survivors with trisomy 9 mosaicism. *American Journal of Medical Genetics Part A*, 155A(5), 1033-1039. DOI 10.1002/ajmg.a.33928 (n=14)

Highlighting collaborations: Martina Biagi

- Martina Biagi and Io Sono project (2020)
- Acceptance of Io Sono documentary at multiple film festivals across Europe
Interdoc Festival,
Bridges International Film Festival, Constanta International Film Festival, Golden Tree International Film Festival, and Bosifest



The E. WE Foundation

- Sarita and Kareem Edwards founded The E. WE Foundation; parents of Elijah, diagnosed with trisomy 18.
- Began in January 2021; asked to join Advisory Board in April 2021
- Participated in advocacy conference (June 2021)
- Podcast in March 2023:
https://www.youtube.com/watch?v=109-7fjL_LM
Ep 77. [#Trisomy](#) Awareness Month!
Hear from E.WE Foundation Partners
- Member of The Trisomy 18 Coalition



More information at <https://theewefoundation.org/>



E.WE Foundation
A Healthcare Advocacy Organization

SOFT USA



- Ongoing support with recruitment
- Participation in in person and virtual conferences since 2009
- Call outs on Facebook page
- Member of Medical and Scientific Advisory Board

SOFT UK



- Providing information and resources
- Preparing for virtual SOFT UK conference
 - Video overview of TRIS project
 - Q&A ZOOM session in September 2021
- Professional Adviser
- Assisting with SOFT UK Research page
- Podcast January 2025 (available at <https://open.spotify.com/episode/6J1DhekJMIgJpxEbURozUX>)
- Contacts with families in the UK

Trisomy Collaborative

- Promote research, education and training, and advocacy to facilitate professional organizations', health systems' and clinicians' capacity to support parents' experience with trisomy 13 and 18, including decision-making about clinical options.
- Initial meeting in Chicago in June 2025 for planning
- More information will be shared at <https://www.seattlechildrens.org/research/centers-programs/bioethics/our-labs/wilfond-lab/research-projects/#trisomy>



Requests from parents, students and professionals

- Range of individuals with direct contact and/or interaction with child(ren) or adult(s) with a rare trisomy condition
- Contacts from around the world including USA, UK, Algeria, Thailand and Netherlands
- Range of topics and information needs (medical, developmental, support, resources)
- Follow up for questions, additional needs etc.

Sample parent requests

- Requests from pregnant moms
 - General information
 - Related to delivery / birth plan development
 - Specific syndrome related concern such as treatment for cardiac defect
- Requests about specific conditions/treatments
 - Anesthesia risks in t18, t13
 - Weight curve in t18, t9m
 - Seizures and low salt metabolism in t13
 - Seizure medications in t18 regarding effectiveness and potential side effects
 - Gastrostomy tube placement in t9m

Requests from professionals

- Researchers at Washington University (St. Louis) regarding natural history and clinical profile for trisomy 18
- Genetic Counselor at NYU Langone-Maternal Fetal Care Center request regarding working with parents with fetus diagnosed with t8m
- Complex care pediatrician and internist at Gillette Children's Hospital in St Paul, MN regarding care for 22-year-old patient with trisomy 13

Requests from professionals continued

- Case manager from Illinois Crisis Prevention Network regarding 24-year-old with trisomy 13 on caseload. Wants to assist family; asking about medical and behavioral resources
- School social worker in Illinois regarding referral for student for autism evaluation diagnosed with trisomy 13 and Turner syndrome
- School Counselor in Michigan regarding 6th grader with trisomy 9 mosaicism; asking about re teen support groups, camps etc.

Responses to requests

- Share TRIS project materials including
 - Article(s) (TRIS project results and TRIS “Library”)
 - Link to informational modules (full t18, full t13, t9m): <https://tris.siu.edu/modules/>
 - Link to case study: <https://tris.siu.edu/case-studies/>
- Search database and/or completed surveys regarding request, compile data and share
- Assist to connect with parent(s)
- Connect with SOFT USA, UK resources including Care book, and surgery registry, and organizations such as NORD, and UNIQUE

Updates in NORD database

- Updated t9m information in fall 2023; available at <https://rarediseases.org/rare-diseases/chromosome-9-trisomy-mosaic/>
- Updated t18 information in winter 2024; available at <https://rarediseases.org/rare-diseases/trisomy-18-syndrome/>

NORD is the National Organization for Rare Disorders (based in the United States)



Ongoing activities

- Processing new enrollees
- Database management
- Website maintenance/new template
- Responses to queries
- Processing survey completions
- Twice a year updates posted on website
- Tracking general media articles (newspaper and TV)
- Continued collaboration with SOFT USA and SOFT UK



and researcher at the National Institutes of Health (NIH)

- **New direction for the project** **TRIS 2.0**



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TRIS project website links:

<http://tris.siu.edu>

<http://web3.coehs.siu.edu/tris/PreEnroll.php>

<https://tris.siu.edu/tris-project-update/>

<https://tris.siu.edu/modules/>

<http://www.coehs.siu.edu/tris/casestudies.html>

<https://tris.siu.edu/publications-presentations/>

