



Updates on Trisomy 9 Mosaic Syndrome (T9M) SOFT Conference July 2025

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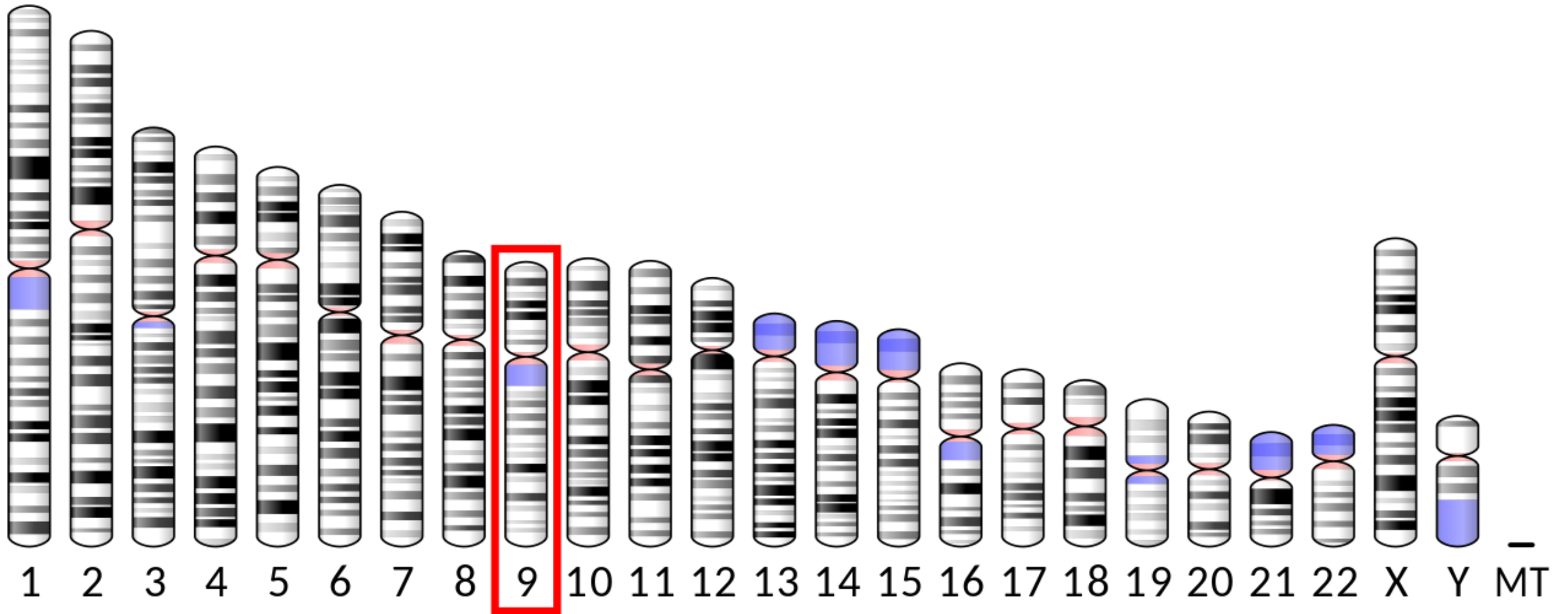
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Introduction

- Welcome and thank you for coming to SOFT!
- Introductions
- Topics for today
 - Background
 - TRIS project introduction/review
 - Medical information review
 - Open Q&A

Chromosomes Lined Up

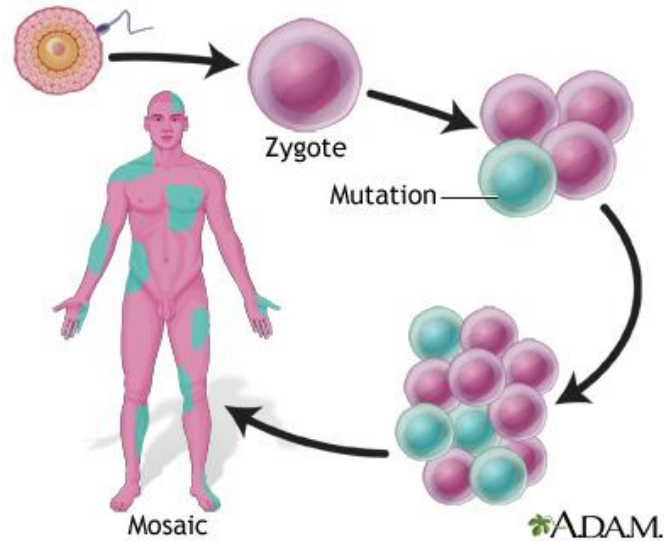


Background: Chromosome 9 Variants

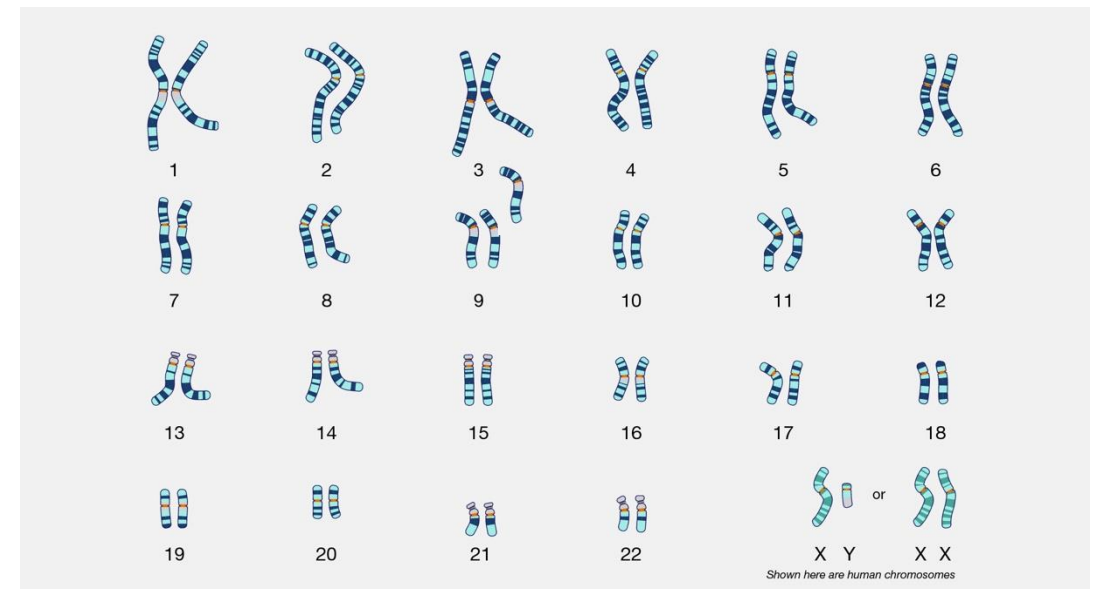
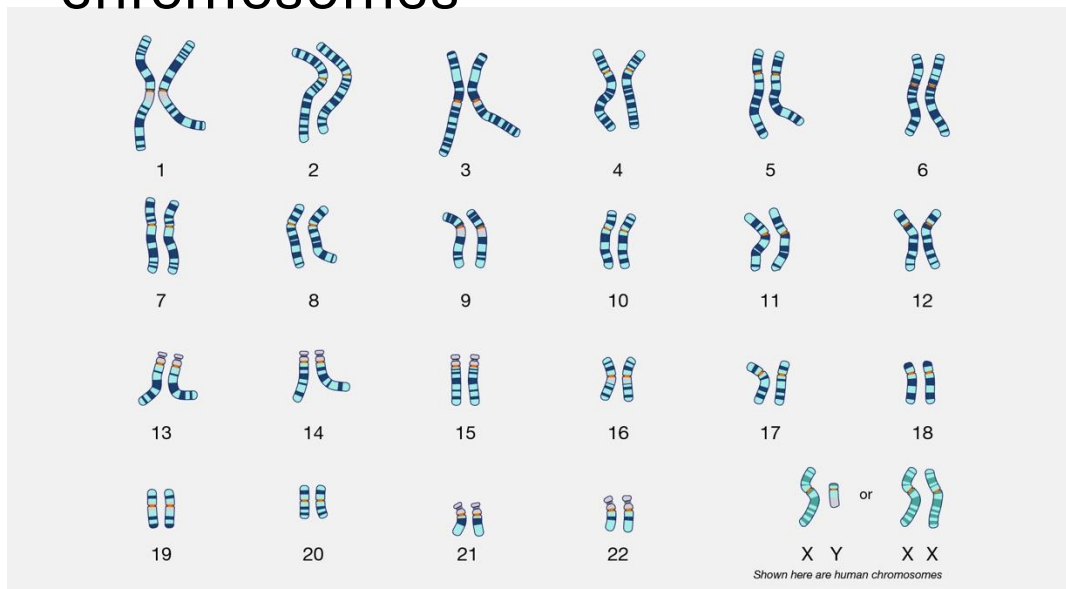
- Trisomy 9 (T9)
 - All cells (building blocks of body) with an additional copy of chromosome 9
- Trisomy 9 Mosaic syndrome (T9M)
 - Presence of some cells with two copies of chromosome 9 and some cells with an additional copy of chromosome 9
- Partial deletions (missing a part) of chromosome 9
- Partial duplications (extra part present) of chromosome 9

T9M

Some cells have the expected number of chromosomes

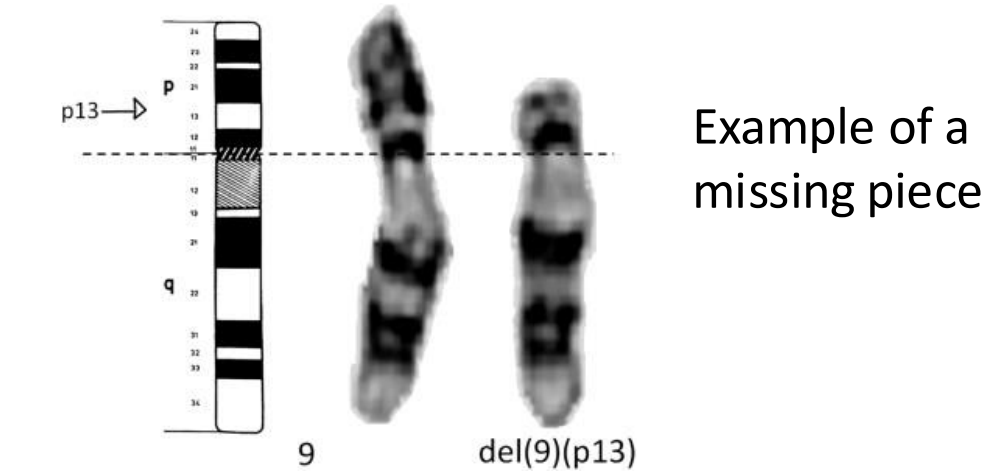


Some cells have an extra copy of chromosome 9

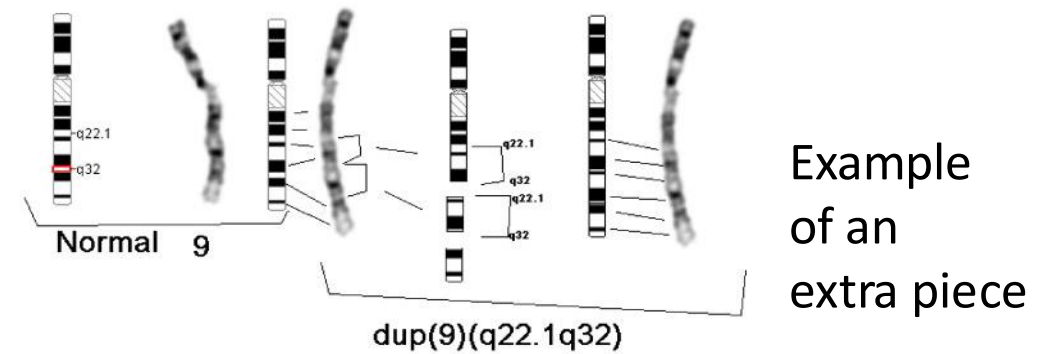


Examples of Chromosome 9 Changes

- Missing and extra pieces of chromosome 9 have been reported
- Some similar features
 - Growth problems
 - Developmental delay
 - Multi-systemic medical issues
 - Different facial features



<https://www.sciencedirect.com/science/article/pii/S102845591400196X>



https://www.researchgate.net/figure/Idiogram-and-partial-karyotype-revealing-duplication-of-9q_fig4_41450690

Earlier T9 and T9M Literature Findings

~40 individuals with T9

>100 individuals with T9M

~30 prenatally diagnosed cases reported
were not confirmed postnatally

Multisystemic medical issues
described

Common craniofacial, cardiac, genitourinary,
skeletal, central nervous system abnormalities

Updating the Literature

- Most initial publications on chromosome 9 variants were smaller reports
- Led to updated publication on T9M with first medical screening guidelines published 2021
- Identified need for a centralized clinical center
- Inspired by and thanks to many of you!

Lurie Chromosome 9 Clinic

- First clinic in the world dedicated to individuals with changes in chromosome 9 was launched May 2024 at Lurie Children's Hospital
- Occurs at least quarterly or more based on need
- Has allowed growth of medical knowledge regarding these conditions
- Clinical spectrum is much broader than what was reported in nearly all areas of health, growth, and development
- Need led to expansion from T9M to include all chromosome variations of Chromosome 9

Lurie Chromosome 9 Clinic

- Individual who live in Illinois may be seen via telemedicine even for initial visits
- Due to state laws, out of state patients must be seen in person at least for the initial visit, then may be seen by telemedicine for a few years
- We have ongoing natural history studies with goal to collect as much history as possible regarding how our patients grow and develop
- Enrollment for studies occurs primarily during clinic visits or events such as SOFT due to time/support limits
- Additional information available online at Lurie's website



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 on children living more than 60 days
 - Modified survey (up to 60 days, miscarriage, termination, stillborn)
 - Follow-up Survey (annual update)
- Responding to requests for medical and/or developmental information for doctor visits, reports etc.
- Outreach (raising awareness, recruitment)
- Dissemination of survey results (conferences, publications)

TRIS project articles on trisomy 9 mosaicism

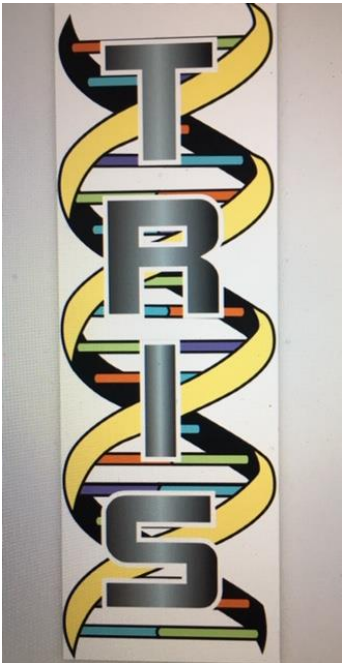
Bruns, D. A. (2025). Presenting characteristics and medical conditions in 67 cases with trisomy 9 mosaicism. *Clinical Genetics*.

<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](https://doi.org/10.1002/ajmg.a.36977)

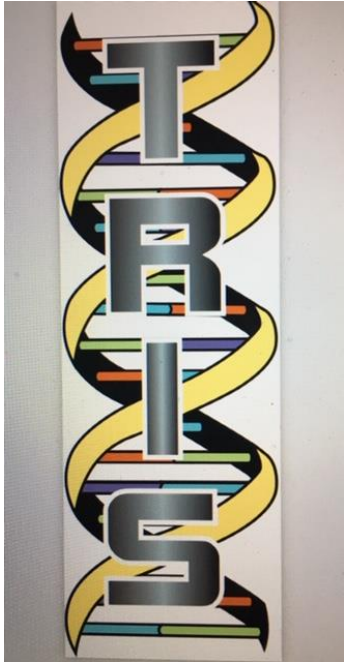
Bruns, D. (2011). Presenting physical characteristics, medical conditions and developmental status of long-term survivors with trisomy 9 mosaicism. *American Journal of Medical Genetics Part A*, 155A(5), 1033-1039. DOI [10.1002/ajmg.a.33928](https://doi.org/10.1002/ajmg.a.33928) (n=14)

Main findings: Physical characteristics



- Affects multiple areas of the body including craniofacial, cardiac, genitourinary, musculoskeletal and neurologic
- Patients often have smaller growth parameters
- A number of unique facial features but not always uniform in presentation

Main findings: Neonatal period



- Feeding and respiratory difficulties are common
- May need support from a feeding tube (mouth, nose, stomach)
- May need extra breathing support (nasal cannula, breathing device called CPAP, ventilator)
- May need extra care in the neonatal ICU depending on findings

Main findings: Range of medical concerns



- Broader than previously described
- Affects a range of organ systems:
 - Eyes
 - Ears
 - Mouth including palate
 - Heart
 - Lungs
 - Feeding
 - Endocrine
 - Genitourinary
 - Muscles/spine/joints
 - Neurologic system and areas of the brain

Main findings: Developmental milestones



- Universally described but varies in terms of severity
- Variation in verbal skills and non-verbal communication
- Communicates with vocalizations, eye gaze, pointing, activate assistive/augmentative communication devices, and some with language (respond to "wh" questions, spontaneous requests, mild to moderate delay)
- Social interactions with preferred adults, siblings, and some peers (school setting, recreation activities etc.)
- Motor skills vary (fine and gross motor)

Update in NORD database

Updated T9M information in fall 2023; available at
<https://rarediseases.org/rare-diseases/chromosome-9-trisomy-mosaic/>

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



Collaboration with Martina Biagi

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



Concerns from parents with a T9M child

- “I think my biggest concern is knowing when enough is enough and letting her pass with dignity. I worry about my ability to let her go. ”
- “...what new medical issues may arise (we had a rough first few years from a medical standpoint) but even more than that is what life will look like for him as an adult. ”
- “Medicaid is also a sore subject right now with all the stuff going on at the national level and in Indiana specifically. Certain legislators are threatening to take away the waivers, which give our kids their Medicaid and if that happens, I know we would go bankrupt trying to pay ”

Ongoing Projects and Future directions

- Working on updating care and management guidelines
 - Collating TRIS study information and Chromosome 9 clinic studies
- Greater outreach to recruit new families to the TRIS project and Lurie natural history studies
- Working on a T9 variation SOFT New and Expectant Parent Resource Guide (T18 and T13 are available at <https://trisomy.org/expectant-parent/#/>)

Acknowledgements

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Questions?



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