

NASCARR

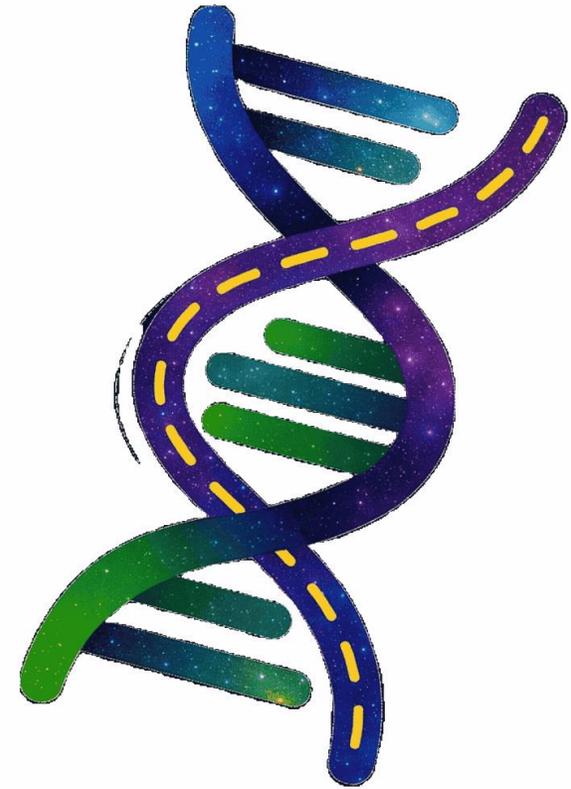
**Network for Advancing Sex Chromosome
Aneuploidy Research Readiness**

**Introducing NASCARR: A new partnership connecting
researchers, families, AXYS, advocacy groups, and the
NIH to advance research
for X&Y Chromosome Variations**

AXYS Webinar March 23, 2026

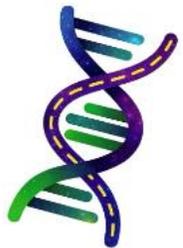
Today's Agenda

- Introducing NASCARR
 - SCA & Research Readiness
 - NIH program (RDCRN)
 - NASCARR Cores & Committees
 - Patient Advocacy Groups (including AXYS)
- INSIGHTS & GALAXY Registries
 - History & Context
 - GALAXY Dashboard
 - Future Goals
- Community Involvement



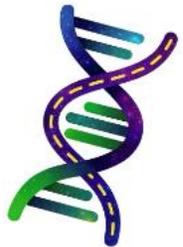
Breaking it down...

- **N**etwork
for
- **A**dvancing
- **S**ex
- **C**hromosome
- **A**neuploidy
- **R**esearch
- **R**eadiness



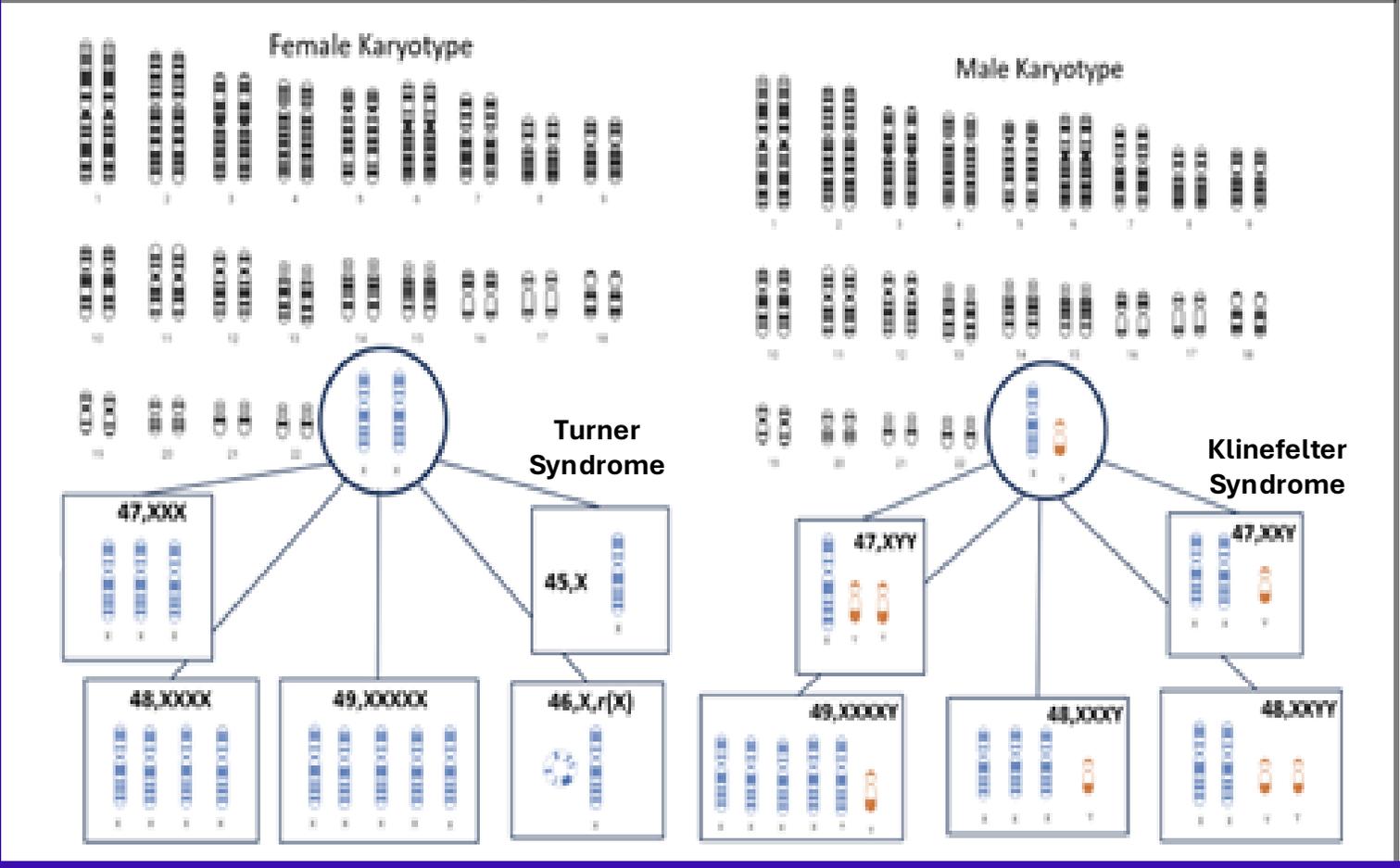
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- Network for
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- Aneuploidy
- Research
- Readiness



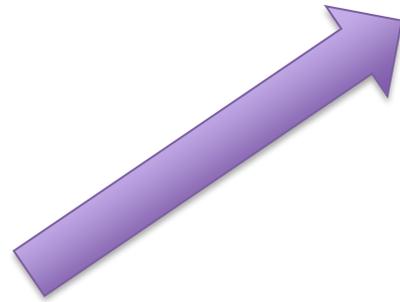
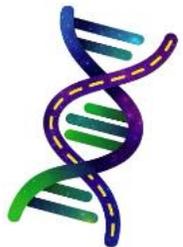
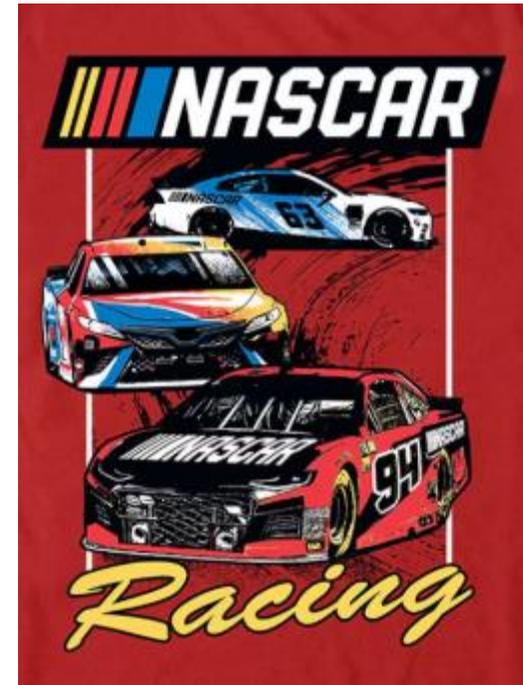
Aneuploidy = Atypical number of chromosomes

SCA = Variations with atypical number of sex chromosomes



Breaking it down...

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- Readiness



Breaking it down...

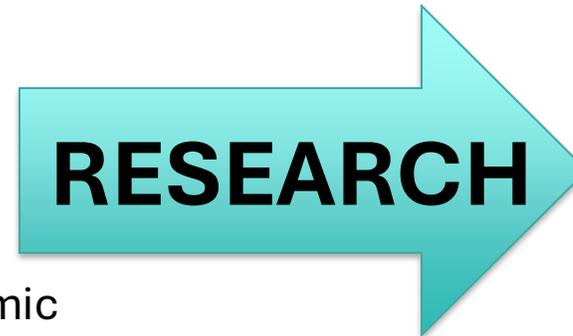
• Network
for

- **Advancing**
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- **Chromosome**
- **Aneuploidy**
- **Research**
- **Readiness**



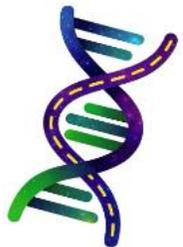
Problem Conditions

Hormonal
Fertility
Fatigue
Other Medical
Developmental
Language
Learning / Academic
Executive Functioning
Social - Emotional
Quality of Life



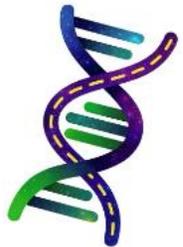
Evidence-Based Treatments

Medical Care Guidelines
Best hormone options
Medication options
Fertility Interventions
Language intervention
Learning strategies
Psychological Therapy
Approaches
Genetic approaches
....and many more

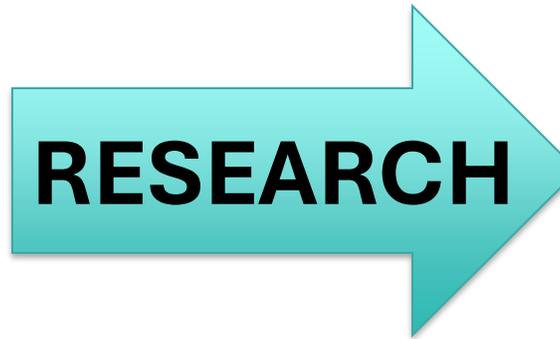


Breaking it down...

- Network for
- Advancing
- Sex
- Chromosome
- Aneuploidy
- Research
- Readiness

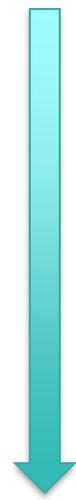


Problem Conditions



Evidence-Based Treatments

RESEARCH READINESS

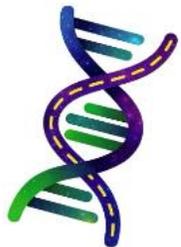


- Researchers and Research Participants
- Background research on topic and intervention
- Best tools to measure change / improvement
- Pilot studies

CLINICAL TRIALS / TREATMENTS

Breaking it down...

- Network for Advancing Sex Chromosome Aneuploidy Research Readiness



Academic Medical Centers



ADVOCACY GROUP PARTNERS

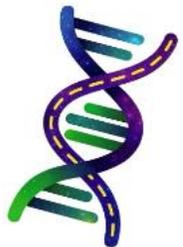
- Association for X and Y Chromosome Variations
- Turner Syndrome Global Alliance
- The XYY Project
- Chromodiversity
- Living with XXY
- TS Foundation
- Turner Syndrome Society of the US

NIH National Center for Advancing Translational Sciences

RARE DISEASES
CLINICAL RESEARCH NETWORK

Breaking it down...

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- **C**hromosome
- **A**neuploidy
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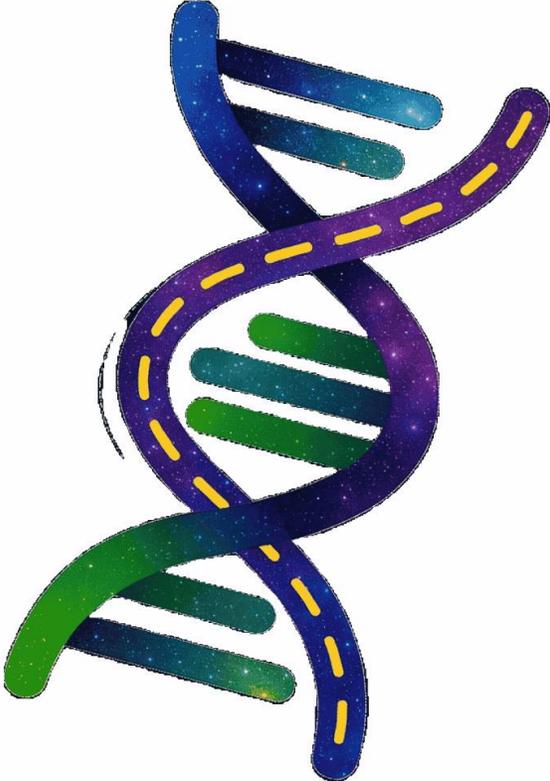
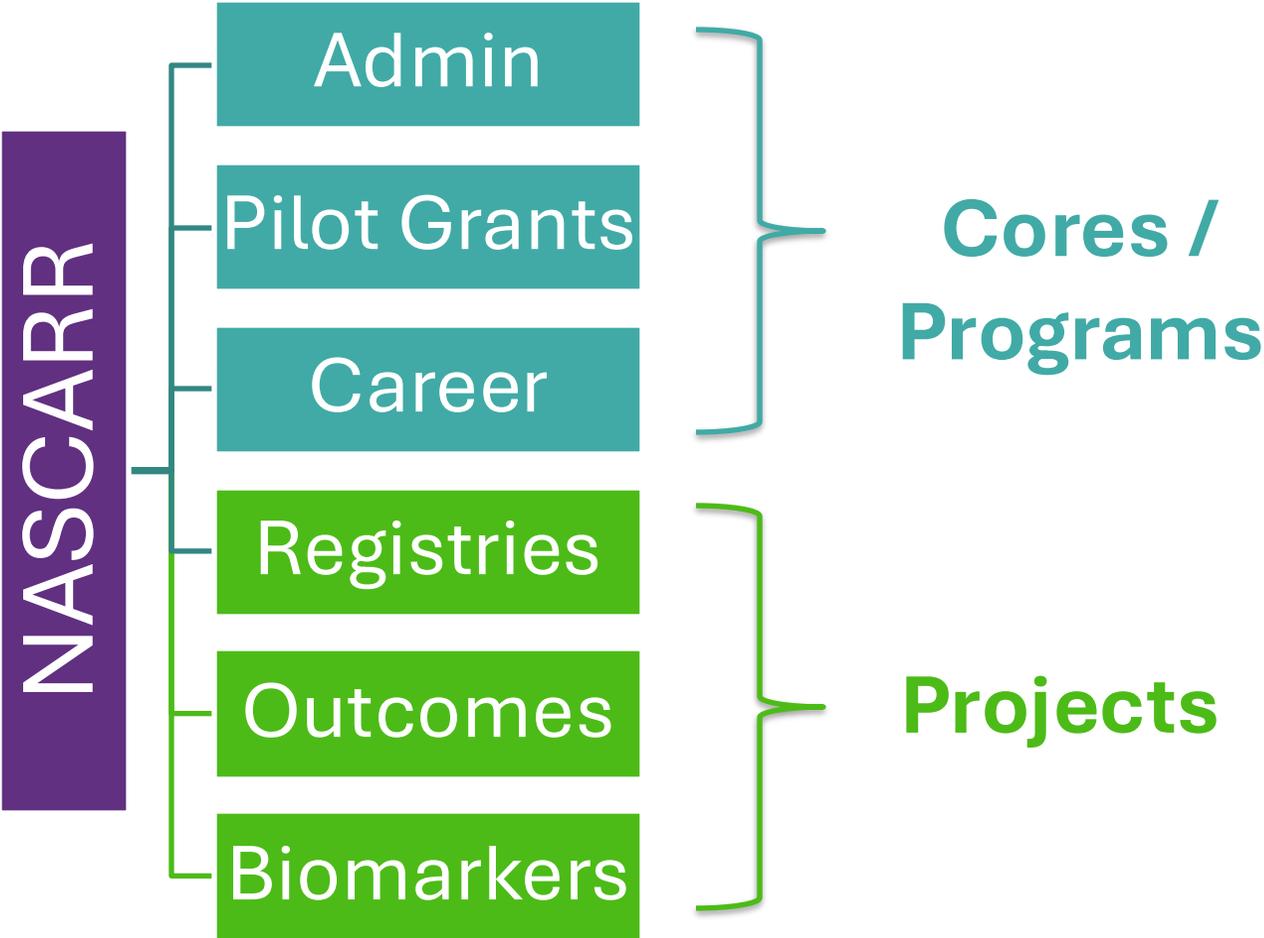
Breaking it down...

NASCARR

**Network for Advancing Sex Chromosome
Aneuploidy Research Readiness**



NASCARR Structure



<https://nascarr.rarediseasesnetwork.org/>



Member Login



[Our Research](#)

[Patients and Families](#)

[Researchers & Clinicians](#)

[Early Stage Investigators](#)

[News and Events](#)

[About Us](#)

An NIH-Funded Rare Diseases Clinical Research Network Consortium



A collaborative network for advancing research in sex chromosome aneuploidies

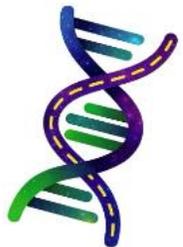
[Learn More](#)



Administrative Core



- Oversight of all Cores and Projects
 - Meetings
 - Collaboration with the NIH / DMCC
 - Committees
 - Communication & Dissemination Committee
 - Partnership Engagement Committee
 - RDCRN Principal Investigator Committee
 - Patient Advocacy Group Committee (AXYS)
 - Budgets



NASCARR RESEARCH SUPPORT TEAM





Pilot Grants Program



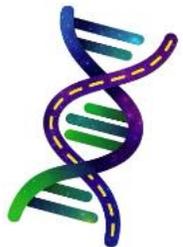
GOAL:

Support research in SCA by funding pilot studies

We can fund 2-3 grants of \$10-15K per year

First year: 12 applications

Award announcements in May 2026

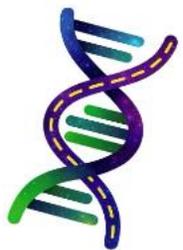


Career Enhancement Program

GOAL: Train the next generation of SCA researchers

Different levels of training / support:

- Summer internship (Undergraduate / Graduate level)
- Medical school rotation in rare disease
- Post-Doctoral Fellowship (Intense research training year)
- Junior Faculty Training and Support



Career Enhancement Core

Career Enhancement Post-Doctoral Fellowship



Application Deadline: March 15th, 2026

The **Network for Advancing Sex Chromosome Aneuploidy Research Readiness (NASCARR)** Career Enhancement Core is committed to growing the next generation of sex chromosome aneuploidy (SCA) clinician-scientists by building a robust clinical research training program and developing high-quality educational resources. **Applications for the Post-Doctoral Research Fellowship are now open.**

The NASCARR fellowship supports a 12-month, mentored research experience focused on SCAs at one or more NASCARR sites. Fellows will engage in hands-on research, attend the national RDCRN Annual Meeting and the NASCARR Annual Symposium, and have access to NASCARR and RDCRN Early Career resources. Prospective applicants should develop a research and training plan in partnership with a NASCARR Senior Faculty mentor that clearly aligns with the program's overarching goal of cultivating independent SCA researchers. Fellows are expected to dedicate a minimum of 50% effort to fellowship-related research and career development. The program welcomes applicants from all disciplines relevant to SCA care, including translational research, molecular biology, clinical genetics, epidemiology, and public health professions. Research in clinical trial readiness will be a priority.

Fellowship Guidelines:

- NASCARR Fellowship stipends may be used to fund travel, fringe benefits, and professional development.
- Awards are for 12 months and are awarded on a competitive basis and contingent upon satisfactory progress and available funds.
- Fellows are expected to attend the RDCRN Annual Meeting in Washington, D.C. in February 2027. Travel and accommodations will be covered.
- Fellows must present work-in-progress at the annual NASCARR symposium September 28-30, 2026 in Aurora, CO. Travel and accommodations will be covered.
- For projects involving human subjects or animals, IRB or IACUC approval must be provided before a subcontract can be executed.
- Awardees must acknowledge NASCARR grant support (U54HD12158) in all manuscripts resulting from work conducted during the fellowship period.



NASCARR Summer Internship Program



Application Deadline: March 2nd, 2026

The **Network for Advancing Sex Chromosome Aneuploidy Research Readiness (NASCARR)** is pleased to support a Research Internship for trainees interested in learning more about sex chromosome aneuploidies and pediatric clinical research.

Sex chromosome aneuploidies are genetic conditions in which an extra or missing X or Y chromosome is present. 1 in 400 babies are born with an extra or missing sex chromosome, but only a handful of centers around the world are conducting research to learn more about these conditions and how best to care for them. These centers apply multiple methodologies (retrospective and prospective observational studies, translational science, intervention trials, secondary data analyses, surveys, qualitative methods, etc.) to answer important clinical questions with the goal of improving patient outcomes in these genetic conditions. Major research endeavors these centers are supporting include the Inspiring New Science in Guiding Healthcare in Longitudinal Analysis in X & Y chromosome aneuploidies, and blood samples from patients with sex chromosome aneuploidies.

Applicants are encouraged to apply to the Research eXpo at the eXtraOrdinary Kids Research eXpo.

Stipend: \$3,800 stipend will be provided for successful applicants selected for the internship. Housing and transportation are the responsibility of the intern.

Location: This internship will largely be in-person. Several sites are available:

- Children's Hospital Colorado / University of Colorado
- Children's National
- Children's Hospital of Philadelphia

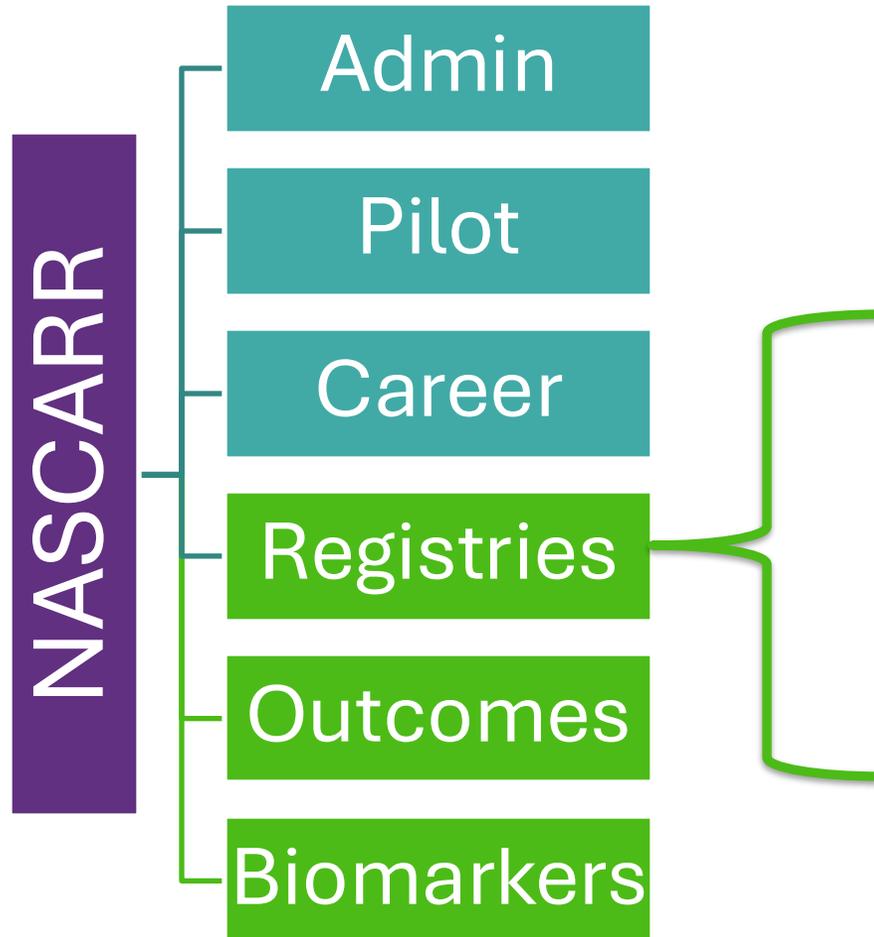
Excellent response to our first call
Reviewing applications now





NASCARR

Network for Advancing Sex Chromosome
Aneuploidy Research Readiness



NASCARR Registries: History & Context

INSIGHTS

- Launched in **2019** with support from Turner Syndrome Global Alliance
- **Population:** Turner Syndrome only



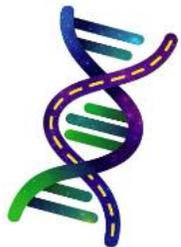
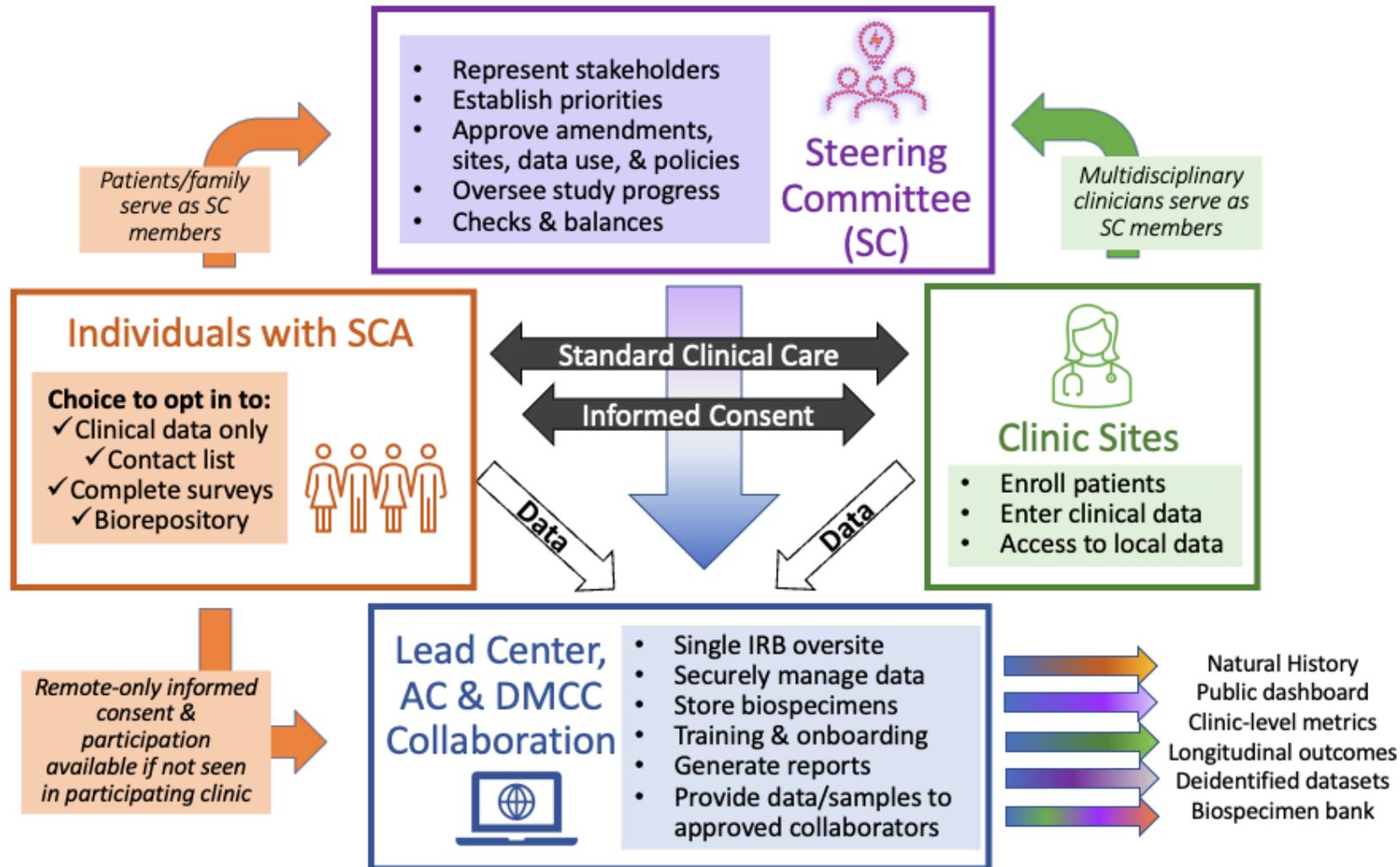
GALAXY

- Launched in **2020** with support from AXYS & Living with XXY
- **Population:** Any X&Y variation

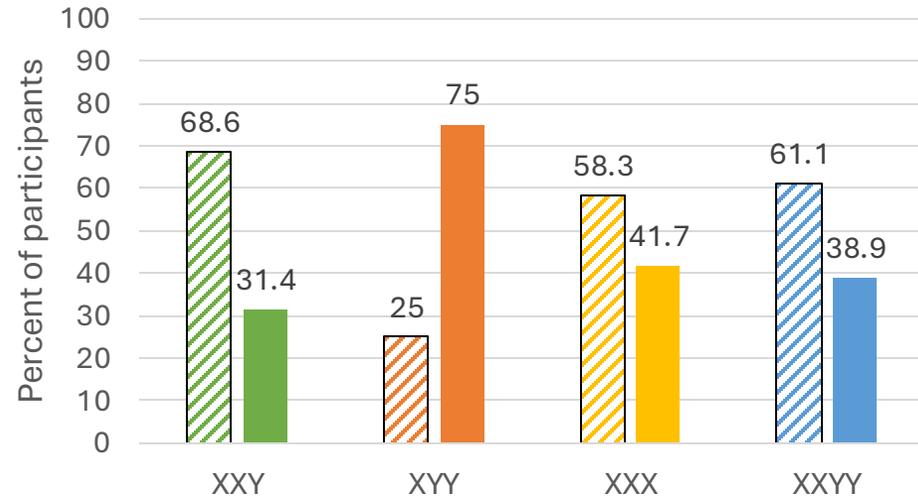


Both are now funded by NASCARR as “Project 1”

NASCARR Registries: Big Picture

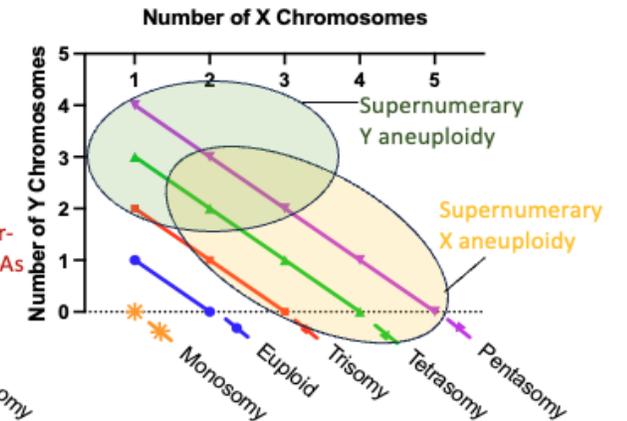
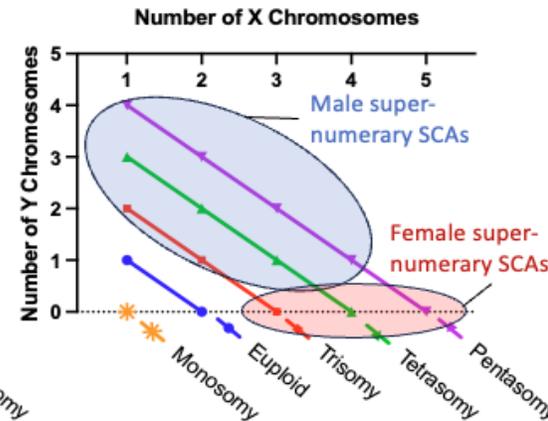
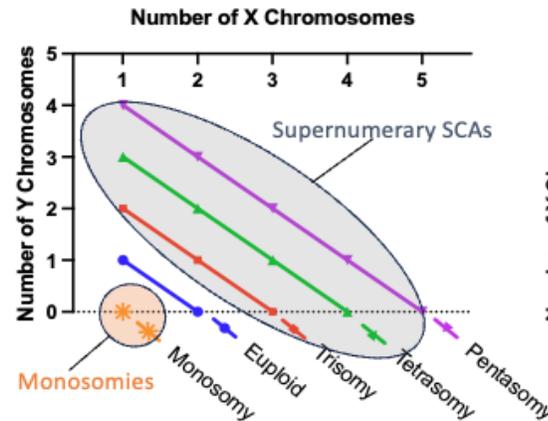
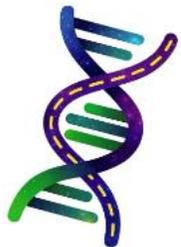


NASCARR Registries: What is the Goal?



Accurate numbers of who is affected by what
 See similarities and differences
 Track changes over time
 Resource for future studies
 Minimize resources and maximize benefits

Complex analyses:
 we need hundreds
 in each group

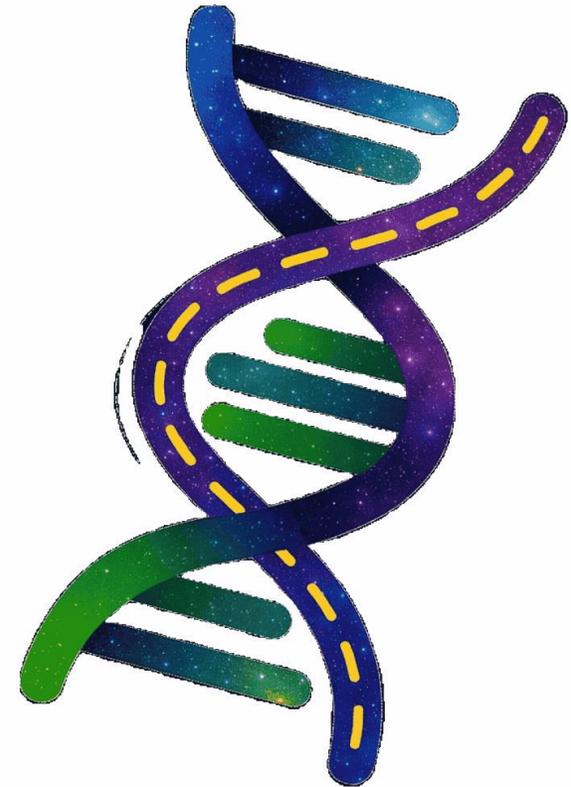


NASCARR Registries: What do I do?

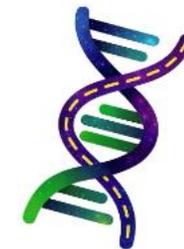
- Medical Record access for research
- List for new study opportunities (optional)
- Surveys (optional)
- Blood sample (optional)



← Consent Form



NASCARR Registries: Progress to Date



- 17 US sites
- 874 individuals enrolled
- 3 publications



<https://redcap.link/INSIGHTS-dashboard>

- 5 US sites
- 595 individuals enrolled
- 3 publications



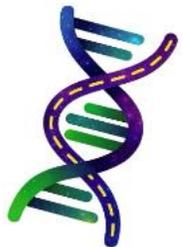
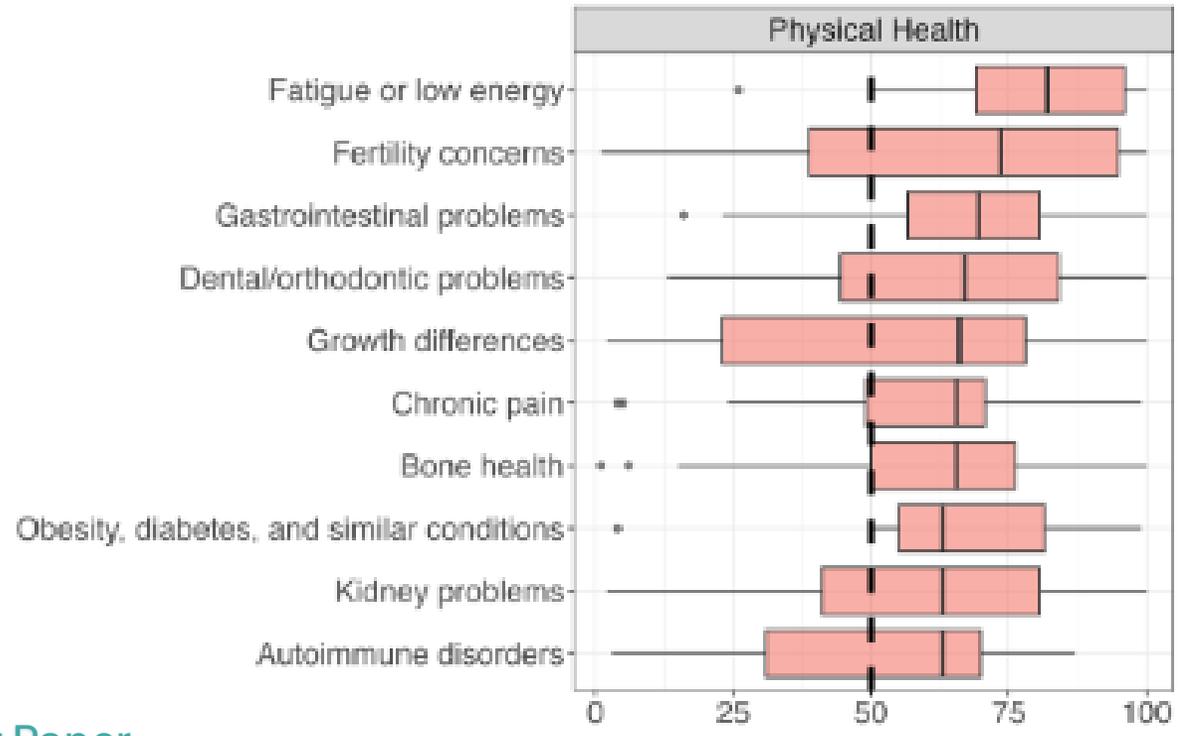
<https://redcap.link/GALAXY-dashboard>

NASCARR Registries: Research Priorities

- Survey developed by the Steering Committees
- >300 participants (48%) completed the survey
 - All SCA conditions
 - Caregivers and self-advocates
- “How important is research on...”



← Full Text Paper



Research Priorities

Not one clear priority

Some topics are of similar importance across all conditions:

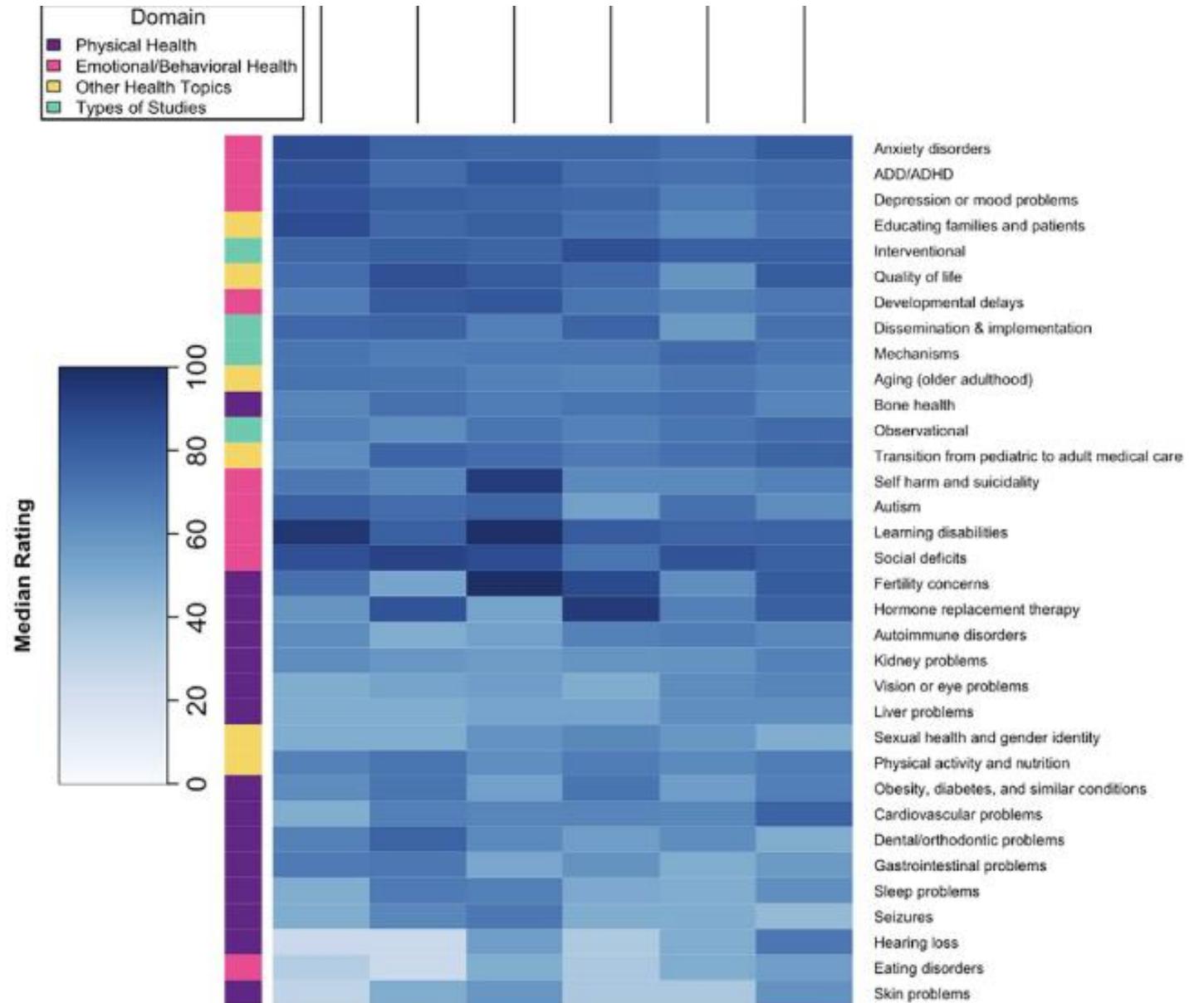
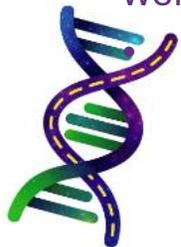
- Anxiety, ADHD, depression

Some topics were more important in 1-2 conditions than the others:

- Hearing loss, eating disorders, skin problems

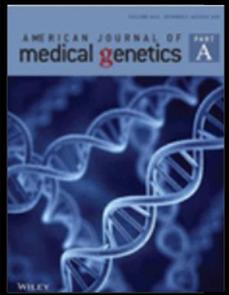
Most topics were similarly important to caregivers and self-advocates, but a couple were more important to caregivers:

Seizures, fertility, self-sufficiency, child development



XXYY Medications

- Research paper and family guide on behavior and mental health medications based on participants with XXYY

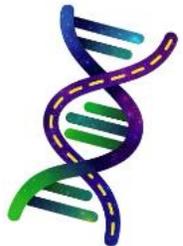


ORIGINAL ARTICLE

Psychotropic Medication Use in 48,XXYY Syndrome

Joanna Dreyer¹ | Susan Howell² | Samantha Bothwell² | Kayla Mollison² | Alexandra Carl² | Karl Swenson² | Shanlee Davis² | Gail Decker³ | GALAXY consortia | Nicole Tartaglia²

¹Developmental and Behavioral Pediatrics, Children's Hospital Colorado, Aurora, Colorado, USA | ²eXtraordinary Kids Clinic, Children's Hospital Colorado, Aurora, Colorado, USA | ³The XXYY Project, Apison, TN, USA



MEDICATIONS FOR BEHAVIOR & MENTAL HEALTH IN 48,XXYY SYNDROME

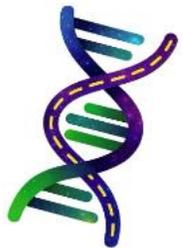
A Medication Guide and Toolkit for Parents & Caregivers of those with XXYY



FUNDED BY THE XXYY PROJECT

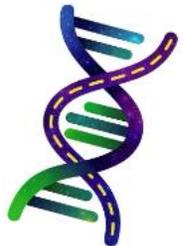
NASCARR Registries: 5 Year Plans

- Increase the number (>2000) and representation of participants (i.e. adults, rarer karyotypes)
- Improve the quality of data in the registry
- Comparisons: to the typical population, between SCAs, and over time
- Expanding the biobank
 - Will be used by “Project 3” to study biomarkers



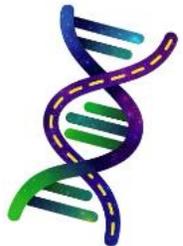
Frequently Asked Questions

- How is GALAXY changing given the NASCARR Funding?
 - Not much. We developed GALAXY anticipating (hoping) for future NIH funding. The protocol and consent forms already included most of the NIH requirements. Additional required language is being added.
 - Data will now be stored in the RDCRN DMCC REDCap instead of the CU REDCap. This change is administrative. All the same protections will remain in place.
- Who owns the GALAXY data?
 - Technically, the University of Colorado “owns” the data in GALAXY
 - Governance of data use still relies on the Steering Committees
 - We will continue to honor that GALAXY is a collaboration between academic and community organizations



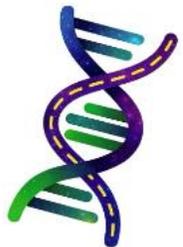
Frequently Asked Questions

- How can I join GALAXY if I am not seen at a clinic?
 - www.galaxyregistry.org → Enroll
 - You must have your genetic test results and/or give permission for us to get these results
- I do not know if I am enrolled in GALAXY, how do I check?
 - Email galaxy@ucdenver.edu to ask about your enrollment status.
- How do I change my choices for the optional procedures?
 - Email galaxy@ucdenver.edu and we will send you a new consent form. *However, you can only enroll in the biorepository in person.*



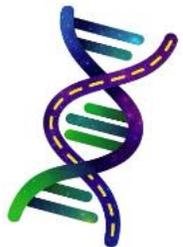
Frequently Asked Questions

- I am already enrolled in GALAXY, do I need to do anything now that GALAXY is under the NASCARR umbrella?
 - No, you do not need to do anything extra right now.
 - We are updating the protocol and consent forms, but this should not require reconsent or other action.
 - All participants will receive an email detailing the consent updates with the option to opt-out of participation if this no longer aligns with your goals.
 - Please respond to surveys you are eligible for! Survey response rate matters.

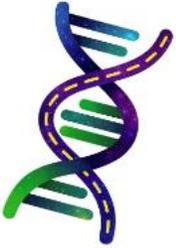


Frequently Asked Questions

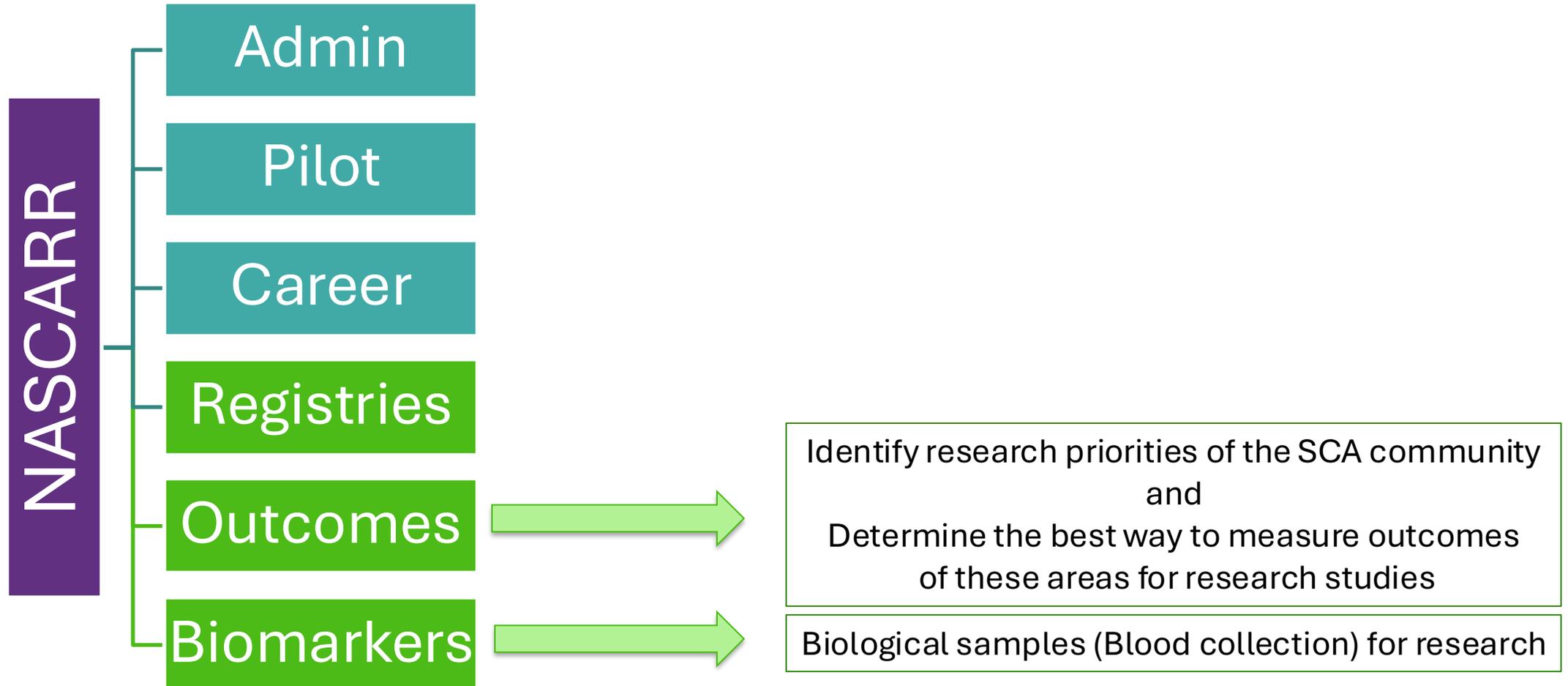
- How do you keep my data safe?
 - Rigorous security protections following all federal, state and local rules to protect your privacy. Data are stored in a password protected database called REDCap only accessible to research team members with appropriate training. Each participant is assigned a unique ID.
- I am not OK with this change to include GALAXY under the NASCARR umbrella, how do I withdraw?
 - You will soon receive an email notifying you that data within GALAXY will be transferring to the RDCRN DMCC. There will be an “opt-out” link.
 - You can withdraw at any time by emailing galaxy@ucdenver.edu. You will be sent a survey to indicate your preferences for existing data/biospecimens.



GALAXY Steering Committee



Dr. Alan Rogol
Dr. Anna Dawczyk
Dr. Armin Raznahan
Dr. David Hong
Gail Decker
Ginnie Cover
Dr. Lilian Cohen
Dr. Shanlee Davis
Steve Gerken
Susan Howell, CGC
Dr. Nicole Tartaglia
Andrew Curry



Opportunities to Engage with NASCARR

- Enroll in GALAXY!
- Join a committee or subcommittee
- Share personal photos or stories for the NASCARR website and/or newsletter
- Participate in Research Projects
 - Stay tuned for announcements

[Email us: NASCARR@UCDenver.edu](mailto:NASCARR@UCDenver.edu)





NASCARR

Network for Advancing Sex Chromosome
Aneuploidy Research Readiness

NASCARR TEAM

Cores



Projects



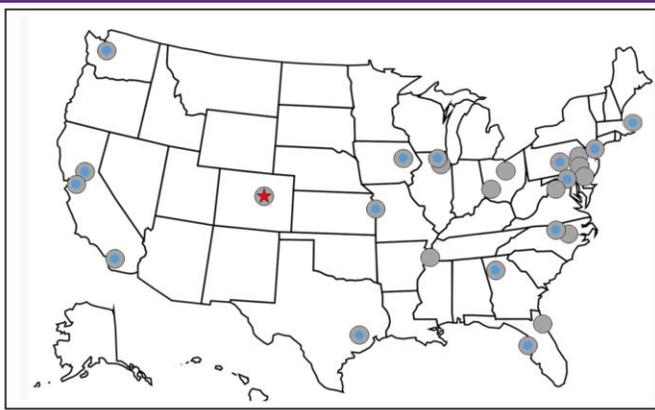


NASCARR

Network for Advancing Sex Chromosome
Aneuploidy Research Readiness

NASCARR TEAM





NASCARR Team

Site Investigators

Dr. Vaneeta Bamba
Dr. Roopa Shankar
Dr. Doris Fadoju
Dr. Catherina Pinnaro
Dr. Melissa Crenshaw
Dr. Kelsie Kelly
Dr. Wendy Brickman
Dr. Elizabeth Debrowski
Dr. Tazim Dowlut McElroy
Dr. Jenn Ladd
Dr. Staci Kallish
Dr. Patricia Fechner
Dr. Karen Klein

Site Investigators

Dr. Jennifer Law
Dr. Siddharth Prakash
Dr. Iris Gutmark-Little
Dr. Maria Vogiaski
Dr. Lilian Cohen
Dr. Courtney Finlayson
Dr. Chijioke Ikomi
Dr. David Hong
Dr. Sejal Shah
Dr. Diane Stafford
Dr. Vicki Elliott
Dr. Leena Nahata

PAG Reps

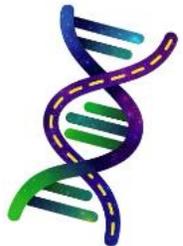
Kelly Ranallo
Liz Rowe
Marybel Good
Gail Decker
Erin Gordon
Elliot Polak
Cindy Scurlock
Ryan Bregante
Kelsey Thomson
Ginnie Cover
Lisa Wolff
Laura Fasciano
Toby Voige
Mariah Hardy

NRST

Sara Deakyne-Davies
Joy Zimmer
Samantha Bothwell
Dr. John Brinton
Dr. Talia Thompson

NIH

Dr. Tiina Urv
Dr. Kathleen Huntzicker
Dr. Melissa Parisi
Dr. Joanne Lumsden



NASCARR Website



Sign up for GALAXY!



GALAXY Dashboard



GALAXY Methods Paper



Research Priorities Paper



Psychopharmacology in XXY

