

AUGUST 2026

ASXL NHS NEWSLETTER

Research updates • enrollment • community

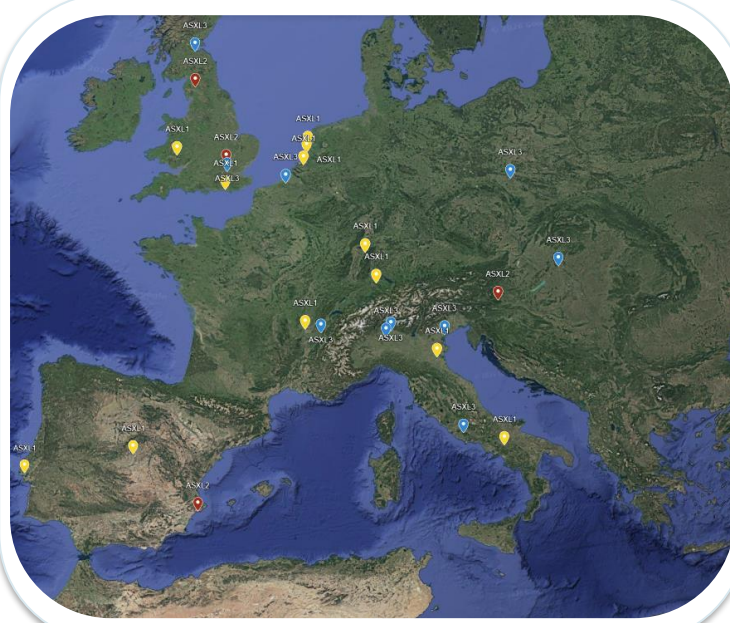
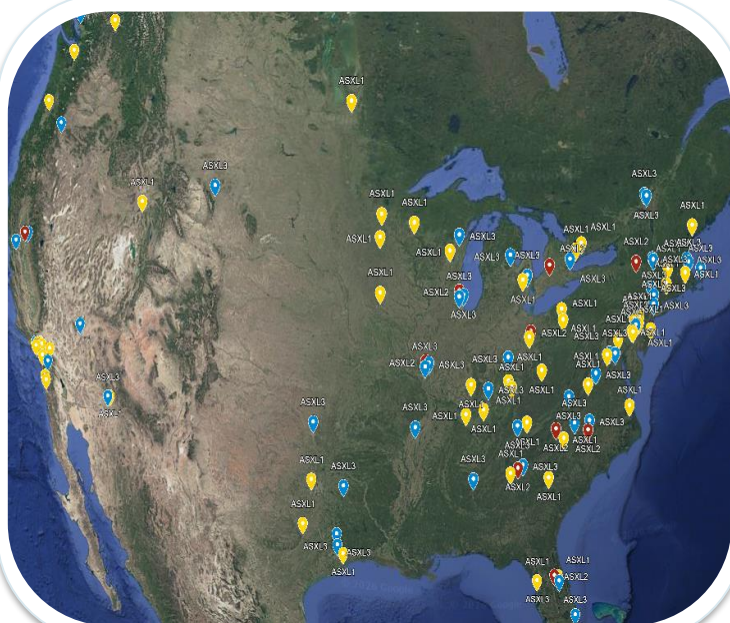
Supporting research and education
for families living with ASXL syndromes
ASXL1 • ASXL2 • ASXL3
ASXL1/ASXL2 • ASXL1/ASXL3 • ASXL2/ASXL3
ASXL1/ASXL2/ASXL3 • ASXL1/ASXL2/ASXL3/ASXL4

ARRE
ASXL RARE RESEARCH ENDOWMENT
arrefoundation.org

BIANCA RUSSELL, MD • NATASHA LUDWIG, PHD • WEN-HANN TAN, MBBS • VANDANA SHASHI, MBBS MD

Our Worldwide Community

The ASXL Natural History Study continues to grow across an international community of families.



200

Families enrolled in NHS

117

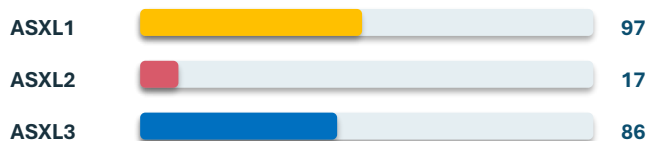
Individuals in REACH Biobank

7

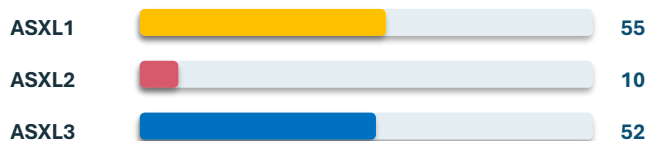
Additional countries noted*

Enrollment by ASXL gene

Natural History Study



REACH Biobank



Help move ASXL research forward

We are actively enrolling participants and collecting blood samples for the REACH Biobank.

Questions about enrollment or participation?

ASXL-CHROMATIN-REGISTRY@mednet.ucla.edu

*Countries not included in the maps: Australia, New Zealand, Canada, Colombia, Brazil, Chile and Argentina.

COMMUNITY & RESEARCH UPDATES

ASXL NHS Newsletter • August 2026

ASXL Family Conference 2026

Thank you for being part of the ASXL Research Symposium and Family Conference!



228
Research visits

116
ALPS assessments

112
Biobank visits

ASXL-Related
Disorders
NATURAL HISTORY STUDY



Insights from Conference Questionnaires

SLEEP (n= 21 (ASXL1= 3, ASXL2 = 2, ASXL3 = 16))

76%

reported at least one
current sleep problem

57%

reported Insomnia

62%

reported current use of sleep
medication/supplement

71%

reported sleep
problems in the past

Sleep concerns may affect nighttime routines, daytime functioning, and family planning — making this an important focus for future natural history research.

COMMUNICATION

Families described multiple communication pathways
(n=18 (ASXL1= 2, ASXL2 = 2, ASXL3 = 14))

- **94%** used aided communication; AAC, iPad, pictures, symbols or word cards
- **90%** often/consistently communicated likes & preferences
- **76%** often/consistently communicated wanting more
- **71%** often/consistently tried to get someone's attention

More than **80%** of parents rated the impact of communication difficulties on quality of life as severe (8-10/10) and **52%** rated communication as the top treatment priority, underscoring the importance of developing and validating communication-focused outcome measures for future research.

GI + FEEDING

Symptoms reported usually/sometimes
(n=18, (ASXL1= 3, ASXL2 = 2, ASXL3 = 16))

Trouble keeping food in mouth	47%
Cough/choke while swallowing	40%
Trouble chewing/biting	40%
Dry heaving / retching	39%
28% currently received some or all nutrition or hydration by tube/IV.	

What We're Learning About Vision

What is CVI?

Cortical visual impairment (CVI) affects how the brain interprets visual information. Our team assessed CVI-related behaviors during the 2024 and 2026 ASXL Family Conferences.

41

participants assessed

80%

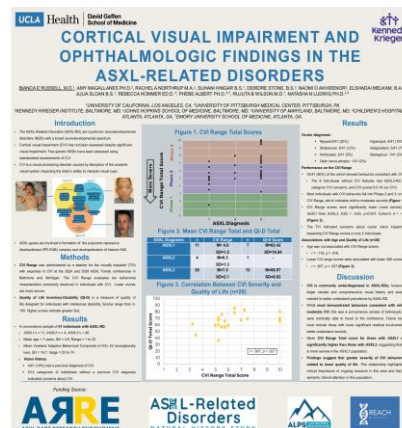
showed behaviors consistent with CVI

10%

had a previous CVI diagnosis

What did we learn?

- Most participants with CVI-related behaviors had mild-to-moderate findings on the CVI Range (0-10).
- Greater CVI severity was associated with lower quality-of-life scores in participants with both measures.
- These findings suggest that visual processing is an important area for future research and clinical care in ASXL-related disorders, with potential implications for understanding daily experiences, functional outcomes, and quality of life.



Poster presented at the 2026 David W. Smith Dysmorphology Workshop

These results describe the 41 participants assessed and do not establish the prevalence of CVI across all individuals with ASXL-related disorders. Larger and more representative studies are needed.

Team Achievements

AUG 2026

Cortical Visual Impairment and Ophthalmologic Findings in the ASXL-Related Disorders

David W. Smith Dysmorphology Workshop • Stevenson, WA

Russell BE, Magallanes A, Northrup RA, Hingar S, et al.

JUN 27-28
2026

Communication in ASXL-RDs: Updates from the ASXL Behavioral Phenotyping Study

ASXL Rare Research Endowment Family Conference • Ann Arbor, MI

Northrup R, Cerf C, Ludwig NN

JUN 25-26
2026

Characterizing Adaptive Functioning in ASXL-Related Disorders

ASXL Rare Research Endowment Research Symposium • Ann Arbor, MI

Northrup RA, Wilson RB, Russell BE, Ludwig NN

JUN 25-26
2026

Characterizing Development in ASXL-Related Disorders

ASXL Rare Research Endowment Scientific Symposium • Ann Arbor, MI

Akhidenor NO, Northrup RA, Wilson RB, Russell BE, Ludwig NN

JUN 25-26
2026

Motor and autism symptoms in ASXL-related disorders.

ASXL Rare Research Endowment Scientific Symposium • Ann Arbor, MI

Wong SC, Northrup R, Tran S, Dinh L, Song J, Le J, Vangala S, McDonald NM, Russell BE, Ludwig N, Wilson RB

Growing ASXL Research Together: Data Sharing

What is a Data Sharing request?

Data sharing helps investigators ask new questions and build on the information families have contributed.

How does a Data-Sharing request work?

Outside researchers follow a structured review process before an approved collaboration begins.



Progress so far

6

data-sharing requests
received

6

institutions
collaborating

WHY THIS MATTERS

Families' contributions can help answer new research questions—while every request is reviewed through a structured process.

How data sharing can accelerate research

- **Connect expertise** → Collaborators can bring specialized clinical, behavioral, genetic, or analytical expertise to ASXL research.
- **Answer new questions** → Existing research data can support additional analyses without asking families to repeat every assessment.
- **Build stronger evidence** → Combining perspectives across institutions can help identify patterns and inform future studies.

Each approved collaboration follows the applicable privacy, ethics, and data-sharing requirements.