

NEWSLETTER



Exciting Update

As of July 2024 we have 544 participants enrolled!

A huge thank you to all the families that have participated in the AS Natural History Study and contributed to this important research!

Here is a breakdown of our participants by age and molecular subtype from 2006 to present

Molecular diagnosis	Age at Last Visit Categories (years)							Total (N=544)
	0-1	2-5	6-12	13-17	18-25	26-45	46+	
Deletion	22	115	122	41	24	26	2	352
Imprinting Defect	0	5	22	6	3	1	0	37
Uniparental Disomy	2	9	32	7	2	2	0	54
UBE3A Mutation	2	24	31	8	12	3	0	80
UPD/ICD	3	1	4	0	3	2	0	13
Abnormal DNA Methylation only	0	2	2	0	3	1	0	8

Recent Publication

Developmental milestones and daily living skills in individuals with Angelman syndrome

What Was Done?

- Study involved 261 AS Natural History Study participants with various molecular subtypes
- Caregivers answered questions about their child's developmental milestones and daily living skills
- Researchers examined typical age ranges and likelihood of skill achievement across molecular subtypes

What Was Learned?

- Individuals with deletion are less likely and slower to acquire skills compared to those without a deletion
- Skills such as sitting and walking with support are likely to be achieved within a narrow age range
- Toileting and feeding skills are less likely to be acquired and have more variability in age of achievement
- Independence in brushing teeth, bathing, and dressing is rarely achieved

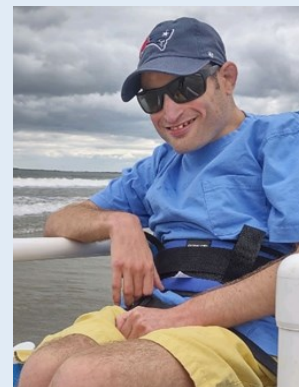
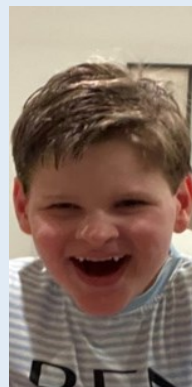
What Does This Mean for Families?

- Achievement of daily living skills and developmental milestones can vary based on molecular subtype
- Teaching daily living skills from an early age through adulthood is important for individuals with AS
- Clinicians and families can use the findings of this study to monitor developmental progress and plan interventions

Citation: Sathwani, A., Powers, S., Wheeler, A., Miller, H., Potter, S. N., Peters, S. U., Bacino, C. A., Skinner, S. A., Wink, L. K., Erickson, C. A., Bird, L. M., & Tan, W. H. (2024). Developmental milestones and daily living skills in individuals with Angelman syndrome. *Journal of neurodevelopmental disorders*, 16(1), 32.



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“Why participate in the AS NHS?”

“The study has linked our family with incredible doctors, researchers, and clinicians that have helped foster a better understanding of our child's condition, raising our advocacy awareness, and connecting us with other Angelman Syndrome families/communities.”

-Mother of 13 yo with deletion

“Since [our daughter] is now 51, we feel as though our experiences and information can make a difference in the lives of other angels. We can also learn from others facing the same daily challenges.”

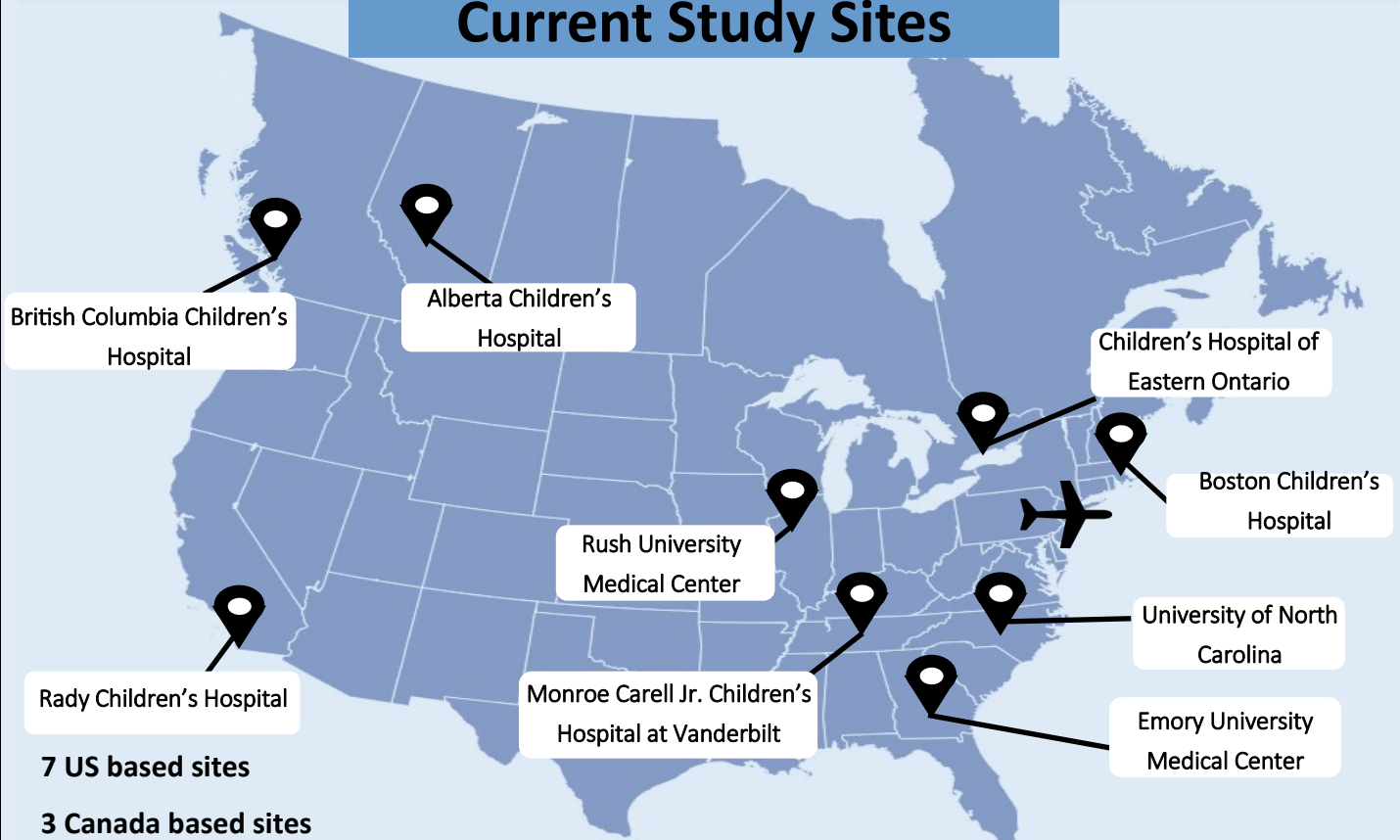
-Mother of 51 yo with deletion

“I received recommendations to take back to the Individual Service Plan team and since they came in writing, from a doctor and team with experience and knowledge about Angelman Syndrome, it was easier for me to get those recommendations implemented.”

-Mother of 42 yo with a non-deletion



Current Study Sites



Please Contact Us to Participate!



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