

LADDER
Database

Linking Angelman & Dup15q Data for Expanded Research



Adults living with Angelman and Dup15q Syndrome

Social media campaign



While the first symptoms of Angelman syndrome and Dup15q syndrome begin in early childhood and most individuals are diagnosed around 1 year of age, individuals with these disorders can live long, full lives. But adults are less frequently represented in research studies like LADDER.

In May, LADDER launched a social media campaign with the goal to increase the number of adults enrolled in LADDER and highlight experiences of caregivers, researchers, and clinicians of adults. We believe by including more adults, researchers can better support affected individuals and their families.

Campaign
snapshots

297

Facebook and
Instagram followers

24

Social media posts

229

Number of total adults
enrolled in LADDER

10%

Percent increase in
adult enrollees

WHY IS IT IMPORTANT TO INCLUDE ADULTS IN RESEARCH STUDIES LIKE LADDER?



Katie Garbarini, Director of the LADDER Learning Network

"As a genetic counselor with expertise in adult genetics, I understand how essential it is to support families with AS and Dup15q over the lifespan. I am passionate about improving our understanding of AS and Dup15q in adults. I look forward to utilizing the LADDER database to answer questions from the community about adulthood with AS and Dup15q. We can use this data to inform resources and initiatives to better support families of adults with AS and Dup15q"



Dr. Tan, clinical geneticist and PI of the Angelman Syndrome Natural History Study at Boston Children's Hospital

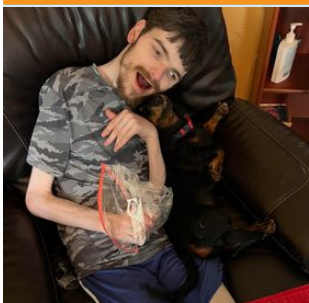
"Much of what we know about the clinical manifestations and complications of Angelman syndrome (AS) stemmed from studies in children and young adults because traditionally, the clinicians who studied Angelman syndrome were trained as pediatricians. Moreover, by the time genetic testing for AS was widely available, many of the adults with AS were no longer seeing physicians who would think of testing them for the syndrome. As such, there is a relative dearth of information on the long-term clinical outcomes of adults with AS"

Snapshots: Quotes from caregivers of affected individuals



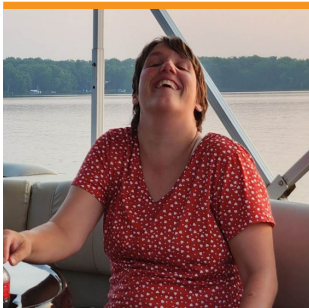
Corrina, age 26 living with Dup15q syndrome

"Many caregivers of adults are skilled advocates and have learned how to get their adult children's needs met...Caregivers also understand the social, emotional and behavioral challenges of adults with dup15q — often times better than the professionals do." - *Nicole, Corrina's mom*



Garrett, age 29 living with Angelman syndrome

"I always say being a caregiver of a special needs adult is like a person that has been practicing a dance with a partner for decades. When we first started and he was a baby, I didn't know the steps, I was always nervous. But after 29 yrs of dancing with Garrett I know his moves." - *Kim, Garrett's mom*



Klara, age 31 living with Dup15q syndrome

"It's important for families with adult dupers to share their stories because it will give others ideas, hope and hopefully love they need to parent a child/adult with Dup15q." - *Michelle, Klara's mom*



Mercedes, age 29, living with Angelman syndrome

"It is a long journey and I can attest that it is worth living, it is very intense but Mechi is our adored treasure. Being in contact with other AS families is very important, I call it my clan. The more information we have and give, the more beneficial for all!!!!" - *Adela, Mercedes' mom*



Troy, age 24, living with Angelman syndrome

"Although our children are not burdens; Angelman Syndrome is burdensome." - *Cassandra, Troy's mom*

Snapshots: Saturday Stats

LADDER SATURDAY STATS

ANGELMAN SYNDROME PARTICIPANTS

143

RANGE: 18 - 50 YEARS

DUP15Q PARTICIPANTS

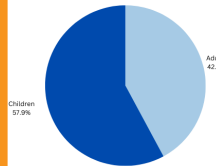
74

RANGE: 18 - 52 YEARS

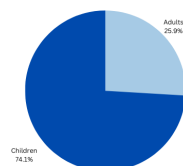


LADDER SATURDAY STATS

Angelman Syndrome: 67 of 159 genetic reports are from adults



Dup15q syndrome: 41 of 158 genetic reports are from adults



LADDER SATURDAY STATS

Angelman syndrome

10% OF CHILDREN
(4 OUT OF 40)

&
26% OF ADULTS
(7 OUT OF 27)

REPORT A DIAGNOSIS OF LGS

Dup15q

32% OF CHILDREN
(11 OUT OF 34)

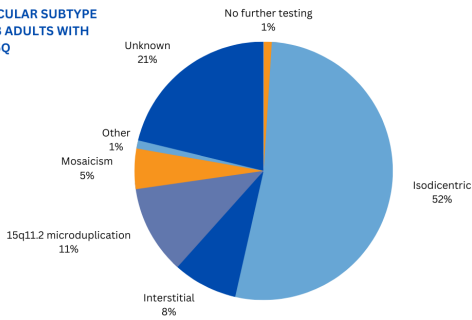
&
32% OF ADULTS
(6 OUT OF 19)

REPORT A DIAGNOSIS OF LGS



LADDER SATURDAY STATS

MOLECULAR SUBTYPE FOR 73 ADULTS WITH DUP15Q



LADDER SATURDAY STATS

PERCENTAGE OF SEIZURE SURVEY DATA FROM ADULT PARTICIPANTS

ANGELMAN SYNDROME PARTICIPANTS

38%

62 OUT OF 162 COMPLETED SURVEYS FROM ADULT PARTICIPANTS

DUP15Q PARTICIPANTS

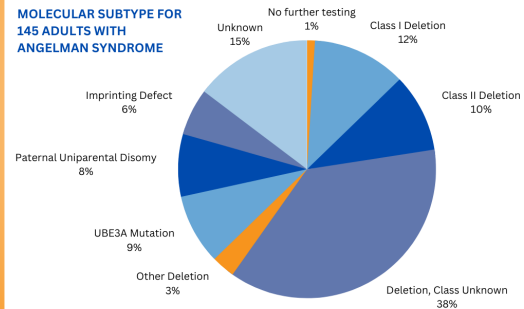
29%

37 OUT OF 138 COMPLETED SURVEYS FROM ADULT PARTICIPANTS



LADDER SATURDAY STATS

MOLECULAR SUBTYPE FOR 145 ADULTS WITH ANGELMAN SYNDROME



Linking Angelman and Dup15q Data for Expanded Research (LADDER) is a database platform that links data collected from multiple sources, such as research studies, registries, caregiver reports, and clinic visits.

By linking these sources of information, LADDER can expand research and accelerate the development of interventions and treatments for individuals with Angelman or Dup15q and their families.

