6 KEY POINTS FROM THE HUWE1 DATA COLLECTION PROGRAM

HUWE1 + RARE-X

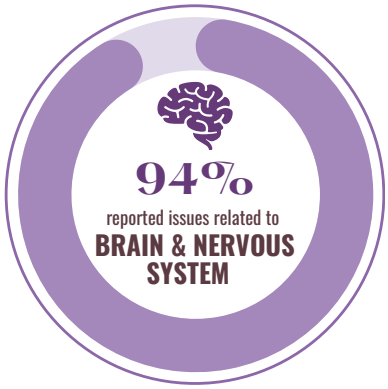
WHAT IS THE HUWE1 RARE-X DATA COLLECTION PROGRAM?

Louie's Huwe, the 501c3 non-profit dedicated to furthering research into HUWE1-related genetic conditions, has partnered with RARE-X to build a Data Collection Program for the HUWE1 community. Patients and caregivers have taken part in **self-report surveys** to help understand symptoms, provide opportunities to participate in research/clinical trials and pave the way to identifying treatments. As of November 2023, we currently have **65 enrolled**, **47 actively taking surveys** and **15 who have provided full genetic details** in reports/clinic notes*. Thank you to everyone who has participated! To learn more visit huwe1.rare-x.org.

LOUIE'S HUWE – FOR HUWE1-RELATED GENETIC CONDITIONS

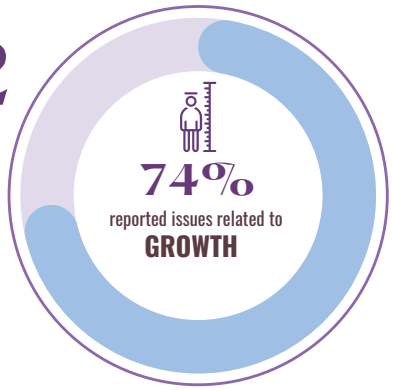
* **Note**: this data is self-reported by participants and the genetic data/variants for each participant have not been verified or classified by researchers or clinicians.

1



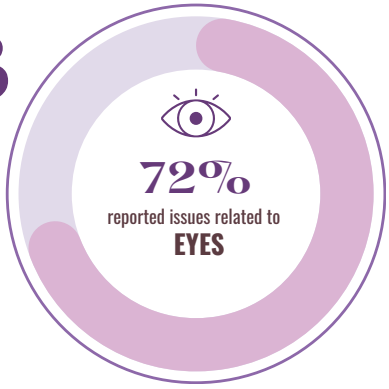
MOST COMMON ISSUES: COGNITIVE IMPAIRMENT, HYPOTONIA, COORDINATION, ABNORMAL EEG, SEIZURES

2



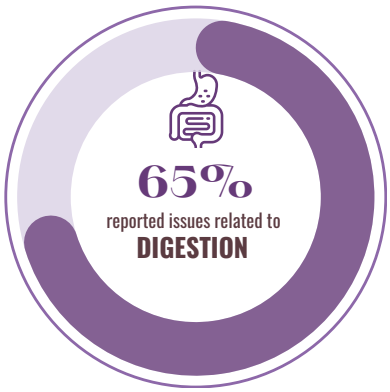
MOST COMMON ISSUES: SHORT STATURE, UNDERGROWTH

3



MOST COMMON ISSUES: ABNORMAL EYE MOVEMENT, FARSIGHTEDNESS, VISUAL IMPAIRMENT

4



MOST COMMON ISSUES: CONSTIPATION, FEEDING DIFFICULTIES

5

MOST COMMON ISSUES: REPETITIVE BEHAVIOUR, UNUSUAL EYE GAZE, DIFFICULTY WITH NONVERBAL BEHAVIOR, AUTISM SPECTRUM, SELF INJURIOUS BEHAVIOR, MELTDOWNS

6 MOST COMMON DEVELOPMENTAL CONCERNS

Motor / Movement / Coordination
Communication (including language)
Delayed or abnormal babbling or speaking
Physical growth
Repetitive behaviours - such as hand flapping or rocking
Abnormal motor development or muscle tone - unusually stiff or floppy
Eating
Did not point, gesture or imitate others