 # 519

INITIAL DATA FROM THE GALAXY REGISTRY: GENERATING ADVANCEMENTS IN LONGITUDINAL ANALYSIS IN X & Y CHROMOSOME VARIATIONS

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Purpose of Study: Sex chromosome aneuploidies (SCAs) are a group of genetic conditions characterized by an atypical number of sex chromosomes. Most SCA research has been limited by small sample sizes, negligible diversity, little SCA community involvement, and a non-longitudinal lens. To address these limitations, we developed the Generating Advancements in Longitudinal Analysis in X & Y Chromosome Variations (GALAXY) Registry, a multicenter clinical data registry that stores the medical data and biospecimens of individuals with SCAs. To better understand registry participant characteristics and to assess current recruitment strategies, we analyzed the demographic and diagnostic information of the participants enrolled within the first 15 months of the GALAXY Registry.

Methods Used: Within its first 15 months, a total of 272 participants enrolled in the GALAXY Registry. Of these 272 enrollees, 168 had complete demographic and diagnostic data (including an available genetic test) and were included in this analysis. Participants were categorized as underrepresented in research if they reported a minority race, ethnicity, gender, language, or payor status (a proxy for socioeconomic status). Diagnostic delay was calculated as the length of time between the date of symptom onset and the date of SCA diagnosis. Descriptive statistics were utilized to express key demographic and diagnostic information.

Summary of Results: The median age of enrollment is 11.5 years, with a range of 0 - 73 years. 71.4% of participants identify as Non-Hispanic White. 38.7% of participants are underrepresented in research, with payor status (19.6%), race (19.0%), and ethnicity (16.7%) being the primary sources of underrepresentation. 54.2% of participants were diagnosed postnatally, with a median diagnosis age of 7.9 years (range: 0 - 48 years) and a median diagnostic delay of 2.6 years (range: 0 - 14 years). Advanced maternal age was the most common reason for prenatal testing (54.7%), while developmental delay before age 3 (34.1%), learning and behavioral concerns (16.5%), and “other” (19.8%) were the most common reasons for postnatal testing. The majority of participants (73.6%) have non-mosaic 47,XXY (Klinefelter syndrome).

Conclusions: GALAXY Registry participants currently have diversity in age and timing of diagnosis, but not in race, ethnicity, or karyotype. Indications for postnatal genetic testing appear largely variable, emphasizing the irregularity of the clinical presentation of SCAs. Future work will focus on increasing enrollment of diverse participants, increasing the number of clinic sites participating in GALAXY, and analyzing how diagnostic and perinatal history vary by karyotype. We will also use the registry to examine the longitudinal course of SCA conditions.