Diversity, equity, inclusion I

Concurrent session

3:15 PM

Thursday, January 20, 2022

#87 STAKEHOLDER ENGAGEMENT EFFORTS TO IMPROVE DIVERSITY IN RARE DISEASE RESEARCH: INSPIRING NEW SCIENCE IN GUIDING HEALTHCARE IN TURNER SYNDROME (INSIGHTS) REGISTRY

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Purpose of Study Turner syndrome (TS) occurs in ~1 in 2,000 females who are born with partial or complete absence of the second sex chromosome. Like many rare disease conditions, most research in TS has been focused on specific features (particularly growth), limited to single centers, included minimal diversity, and lacked community engagement. The Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry was developed to address these limitations.

Methods Used A Steering Committee with stakeholders comprised of researchers, multidisciplinary clinicians, and patient advocates was formed to develop the goals, infrastructure, data collection tools, protocols and engagement strategies for a national, collaborative clinic-based longitudinal registry for individuals with TS. Six institutions with multidisciplinary TS clinics across geographical regions were onboarded as recruitment sites with the goal of >80% of eligible patients enrolling with diversity in age, race, ethnicity, payor status, and timing of diagnosis. The team identified patient-centered multidisciplinary outcomes obtainable through medical records and optional additional study procedures.

Summary of Results To date, 154 participants representing all regional centers have enrolled in InsighTS with an average enrollment rate of 15 per month. The average age at enrollment of 11.9 ± 11 years (range 0–67, 16.9% ˜18 at enroll-ment). 18.5% identify as Hispanic/Latinx ethnicity and racial distribution includes 6.2% Asian, 13.7% Black, 71.9% White, and 11.0% Other Race. TS was identified prenatally in 30.3%of participants. The majority of participants agreed to be contacted for future studies (89%), complete annual surveys (83%) and contribute to the biobank (61%).

Conclusions Stakeholder engagement for the development of a national clinic-based registry for the rare genetic condition of TS has successfully led to a diverse cohort representative of the US population. Additional engagement strategies to increase enrollment while prioritizing diversity are underway.