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What's missing in sex chromosome aneuploidies? Representation and inclusion☆☆☆



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Background

Aneuploidies are any deviation to the normal number of chromosomes (i.e., too many or too few). Males typically have one X and one Y sex chromosome while females usually have two X chromosomes. Klinefelter syndrome (KS, one or more X chromosomes) and Turner syndrome (TS, missing/defective X chromosome) are the most common sex chromosome aneuploidies and often seen in pediatric practices. Both KS and TS cause hypogonadism (low sex steroids), infertility, and are associated with other physical and psychological comorbidities (Dwyer et al., 2015). Moreover, KS and TS are associated with diminished health-related quality of life (HR-QoL). Effective pediatric nursing practice requires a holistic approach; this demands an understanding of the individual's response to their health, not just the pathology of the disease itself. As such, understanding the aspects of HR-QoL impacted by KS/TS is important for guiding clinical decision making and developing pediatric nursing interventions to facilitate health and wellbeing.

Given that KS/TS happen by chance, theoretically, these rates should be similar across racial/ethnic groups. Namely, the rates of KS/TS should be similar among non-Hispanic Whites and Black, Indigenous, People of Color (BIPOC) communities. Currently, it is unknown to what extent HR-QoL has been examined in BIPOC communities affected by KS/TS. To provide culturally-informed care for all patients with KS/TS, it is critical to understand the experiences of BIPOC communities. We conducted a systematic review of the literature on HR-QoL in KS/TS to examine the extent that BIPOC communities have been included and represented in the literature to date.

☆ The Pediatric Endocrinology Nursing Society (PENS) is committed to the development and advancement of nurses in the art and science of pediatric endocrinology nursing and to improve the care of all children with endocrine disorders through the education of the pediatric healthcare community. To aid in achieving that goal, the purpose of the PENS department is to provide up-to-date reviews of topics relevant to the PENS membership and to the general readership of the Journal of Pediatric Nursing

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Methods

We conducted a systematic scoping review of the literature for KS and TS in several databases (MEDLINE, APA PsychInfo, Embase, CINAHL, Cochrane, and grey literature respectively). Briefly, the Boolean literature search used medical subject headings (MeSH terms: KS, TS, quality of life, physical health, physiological, level of independence, social relations, environment, spirituality, faith, personal beliefs) to identify relevant articles. Empirical studies published in English assessing HR-QoL in patients with KS/TS were considered (no restriction on publication date). Following removal of duplicates, study titles/abstracts were screened to identify articles for full text review (Fig. 1). Full text articles were reviewed per inclusion criteria and any disagreements were settled by discussion. Subsequently, patient race/ethnicity demographic data was extracted from included studies for descriptive analysis.

Results

After title, abstract and full text review, the systematic search yielded 55 articles (KS: $n = 24$, TS: $n = 31$) (Fig. 1). Notably, the vast majority (48/55, 87%) of the included studies did not provide self-reported race/ethnicity demographic data. In some cases, studies used country (i.e., Australia, Denmark, Netherlands) as a proxy for reporting race/ethnicity - either stating that nearly all participants identified as white/Caucasian or represented a "typical" social and racial composition of the respective country without any specific detail. To determine percentages of participants from BIPOC communities, data was extracted from five studies on KS ($n = 511$ total participants) (Close, 2011; Close et al., 2015; Liberato et al., 2017; Tartaglia et al., 2010; Turriff et al., 2011). For TS, data was extracted from two studies ($n = 118$ total participants) (Sutton et al., 2005; Thompson et al., 2020). From the seven studies with information available on race/ethnicity, most participants (522/629, 83%) reported as non-Hispanic White (KS: 411/511, 80%; TS: 111/118, 94%). Further, 74/629 (12%) of participants had missing values (i.e., no reported race/ethnicity). These data indicate that KS/TS in BIPOC communities are not represented in the HR-QoL literature (Asian: $n = 7$, 1.4%; Black: $n = 4$, <1%; other: $n = 22$, 4.3%).

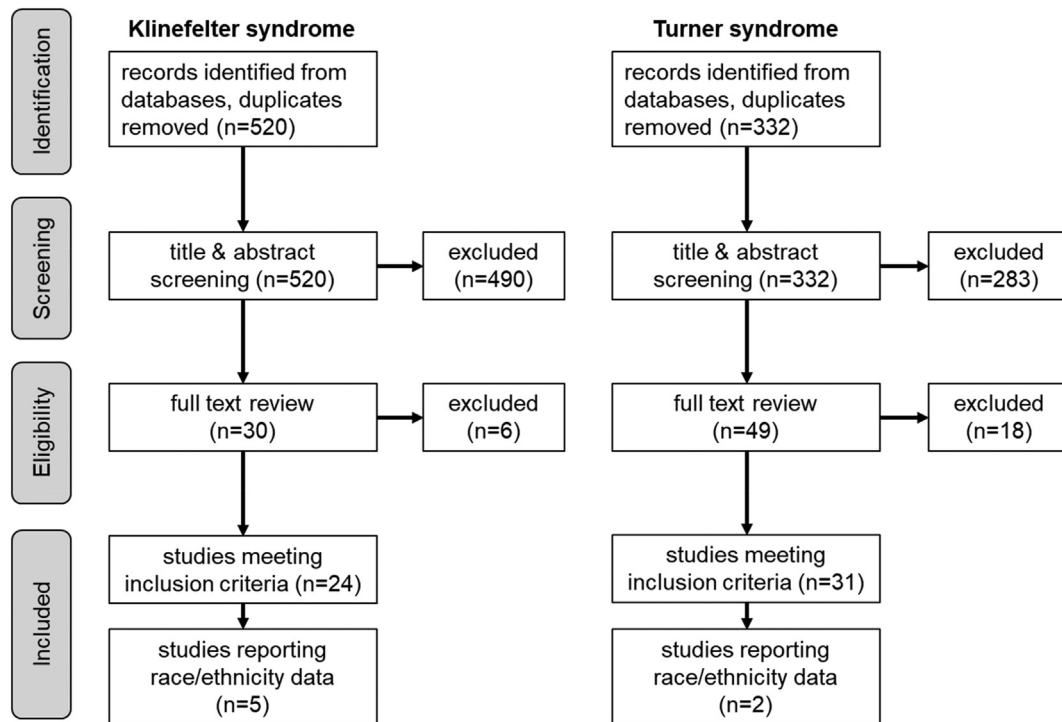


Fig. 1. PRISMA flow diagram depicting study selection process.

Discussion

Herein, we report findings on a systematic scoping review of the literature on HR-QoL in KS/TS. Notably, our findings indicate that patients from BIPOC communities are largely under-represented in studies examining HR-QoL in KS/TS. If one excludes missing values from the denominator, 522/555 (94%) of participants in the seven studies identify as non-Hispanic White – underscoring the significant lack of diversity and inclusion in KS/TS HR-QoL research. Importantly, the lack of evidence for BIPOC communities poses significant barriers to developing person-centered, culturally grounded approaches to improving health and wellbeing of patients with KS/TS. As such, this gap in our understanding has important implications for pediatric nursing practice in caring for patients with KS/TS.

There are a number of potential explanations for the current findings. First, our limited number of identified studies indicate that transparent recording of self-reported race/ethnicity has yet to be widely adopted. Second, BIPOC communities may be distrustful of the healthcare system (and research participation) due to past experiences of discrimination and marginalization. In addition, one should consider the intersection of having a sex chromosome aneuploidy and the economic/social conditions influencing individual/group differences in health status (i.e., social determinants of health). Indeed, gaps in healthcare access may contribute to the observed under-representation of BIPOC communities. Further, much of the research on HR-QoL is conducted in academic medical centers. It is plausible to consider that social determinants of health, structural inequities, and racism may pose barriers to patients with KS/TS attaining care where research is being conducted. Regardless of the root causes, the American Nurses Association has a Code of Ethics that calls on nurses to recognize human dignity regardless of race, culture, creed, sexual orientation, ethnicity, gender, age, experience, or any aspect of identity (*American Nurses Association (ANA), 2020*). Therefore, it is imperative that future clinical, research, and policy practices include BIPOC communities and ensures the inclusion of diverse samples.

Nurses have an ethical mandate to educate, advocate, and collaborate to end systemic racism and promote high quality, equitable nursing

care. To reach this goal, we call on pediatric nurses to advocate and take action in clinical, educational, research, and policy settings. We provide several avenues for pediatric nurses to enact change across diverse settings.

- **Clinical practice:** Nurses can act to oppose and address racism and discrimination in all of its forms by utilizing active listening to engage in and promote deliberate, respectful dialogue across work and community settings. Nurses can also engage in self-awareness to identify their own biases and find ways to address them.
- **Education:** Universities can adopt training programs to build faculty competencies in facilitating discussions on structural inequities and social determinants of health as key drivers of health outcomes. Institutions can encourage and empower faculty to diversify their curriculum by incorporating content and applied case studies into curricula to integrate and embed concepts related to racial/ethnic diversity into nursing practice.
- **Research:** Researchers should adopt clear, transparent reporting of race/ethnicity. Investigators can adopt methods such as community-based participatory methods to engage traditionally underrepresented communities and enhance representative recruitment. Researchers can also focus increased attention on examining the intersectionality of race, social determinants of health, and outcomes for health and wellbeing.
- **Policy:** Nurses can endorse policies and legislative/regulatory initiatives that promote inclusivity and improve the availability of, and access to, affordable healthcare.

In summary, aneuploidies such as KS/TS are outcomes of genetic chance and not related to race/ethnicity. Accordingly, aneuploidies provide a unique lens to examine inclusion and representation of BIPOC communities in research on KS/TS HR-QoL. Our findings suggest that little is known about HR-QoL in KS/TS within BIPOC communities. We posit that action is needed within clinical practice, nursing education, research, and policy arenas to improve our ability to provide comprehensive, holistic pediatric nursing care for BIPOC patients with KS/TS.

While the presented exemplar uses the paradigm of sex chromosome aneuploidies, such efforts may be transferable to other patient populations.

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