Practices of Sickle Cell Disease Genetic Screening and Testing in the Prenatal Population

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Introduction

Genetic screening and testing has only recently become an accessible assessment for genetic risk. Unfortunately, these technologies have been underutilized in minority populations despite their usefulness for predicting diseases like sickle cell disease (SCD), often found in African American and Black and Hispanic populations.[1] We surveyed prenatal patients to understand current choices, beliefs and experiences surrounding genetic screening and testing, specifically for sickle cell disease.

Materials and Methods

In this cross-sectional survey, we collected information from 322 women during prenatal visits from July 2019 through May 2021. Responses to questions about pregnancy screening and testing practices were analyzed for trends to identify barriers to care and education about testing and screening for sickle cell disease. Patients were asked to rate whether they agree or disagree with statements regarding sickle cell health behaviors. We used χ^2 tests to compare categorical variables by self-reported race. Binary logistic regression was used to determine the odds ratios and confidence intervals for each outcome.

Results

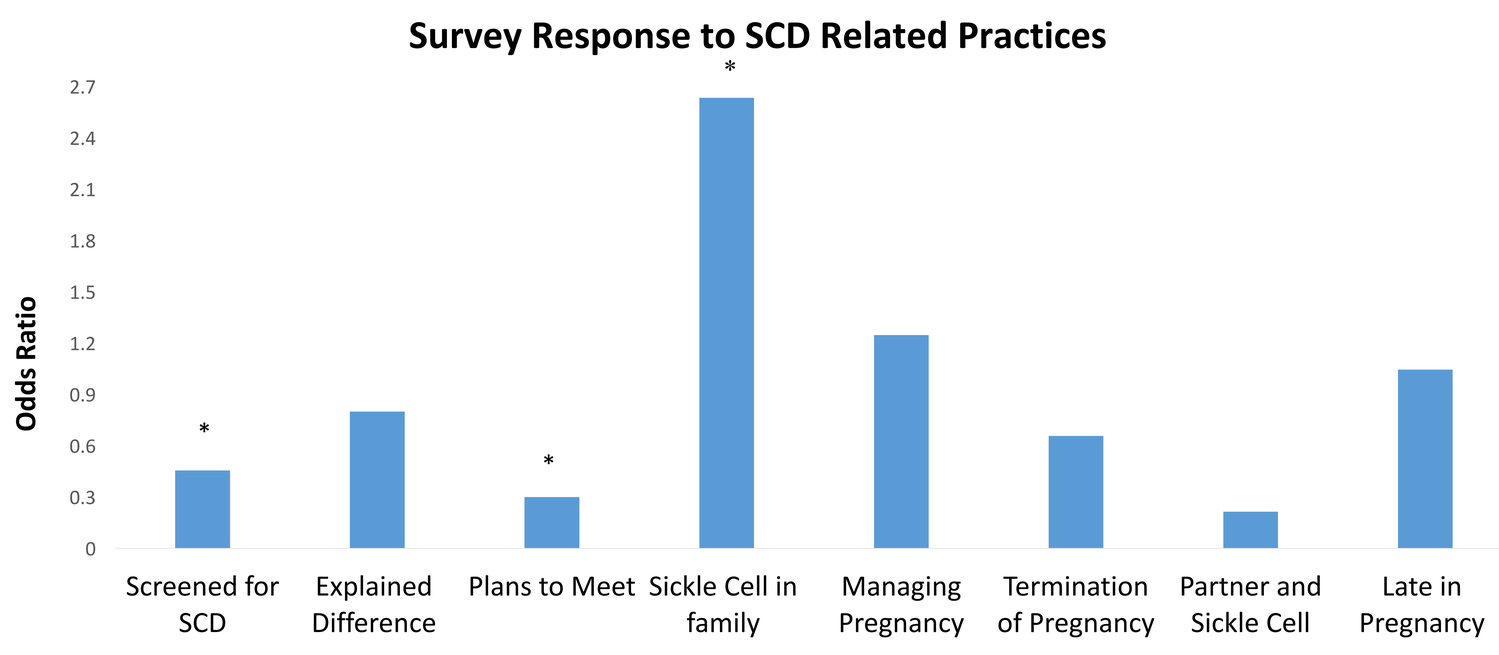

Figure 1: The graph depicts survey responses and odds ratios between White and Black patients. The * depicts statistically significant results from left to right: (p=0.047, OR 95% CI= 0.455 [0.210-0.989]); (p=0.049, OR 95% CI = 0.299 [0.090-0.993]); (p=0.011, OR 95% CI=0.207 [0.081-0.526])

Conclusions

Our findings suggest gaps in screening, testing, and educational efforts between African American/Black and White patients, as well as differences in opinions regarding pregnancy management with a family history or gestational diagnosis of sickle cell disease. Future research should focus on decreasing these healthcare gaps and improving education that address concerns about SCD for relevant populations.

References

1. Reed-Weston AE, Espinal A, Hasar B, Chiuzan C, Lazarin G, Weng C, et al. Choices, attitudes, and experiences of genetic screening in Latino/a and Ashkenazi Jewish individuals. J Community Genet. 2020;11(4):391-403.

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