Genetics Literacy of Sickle Cell Disease: Assessing the Inheritance Knowledge of Young Adults Affected with Sickle Cell Disease

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Sickle cell disease (SCD) is an autosomal recessive hemoglobinopathy that affects an estimated 90,000-100,000 Americans. An additional 3 million Americans are carriers of sickle cell trait (SCT). Although SCD and SCT are relatively common in the United States, the literature reveals a lack of understanding of disease inheritance. Past studies have examined the knowledge of unaffected and affected adults, parents of sickle cell carriers, and unaffected college/ graduate students. However, little is known about the genetics literacy of young adults affected with SCD. Young adults with SCD are a vulnerable population as they transition from pediatric to adult healthcare, and during this period of change and maturation, they are also of childbearing age. We administered a quiz-based questionnaire to assess the genetics knowledge of young adults affected with SCD at Boston Medical Center in Boston, MA. The questionnaire included fundamental SCD/SCT etiology and genetics questions in addition to higher-level SCD inheritance questions. Of 20 participants with SCD surveyed, the average percent of correct responses across all items was 70%. Further analysis revealed a discrepancy in scores between the basic and advanced knowledge questions, with participants faring significantly better with the basic questions compared to the advanced. The results of this investigation suggest that even if affected young adults have a basic knowledge of disease inheritance, they may be unable to translate that knowledge into an advanced understanding of reproductive risk. This disconnect calls for further investigation into how to best educate the SCD population about inheritance and reproductive implications.