Sickle cell trait: Neglected opportunities in the era of genomic medicine

March 6 2014

(Boston)—While acknowledging the potential of genomics to prevent and treat disease, researchers from Boston Medical Center (BMC) and Boston University School of Medicine (BUSM) believe it is long past due to use current scientific data and technical advances to reduce the burden of sickle cell disease (SCD), one of the most common serious single gene disorders. The work, reported as a Viewpoint in this week's *Journal of the American Medical Association* (*JAMA*), highlights the gaps in knowledge and care in terms of SCD and the need to address this issue expeditiously.

The inexpensive identification of the mutated hemoglobin and the technical capacity to screen populations have been known and operative for decades. Sickle Cell Trait (SCT) is estimated to affect 3 million people in the US: approximately 8 percent of African Americans and .5-3 percent of Hispanics. While the screening indications and contexts for life limiting diseases such as cancer, Down syndrome and SCD differ, timely knowledge of genetic vulnerability and genetic counseling are necessary for informed decision making in all screening contexts.

According to the researchers few individuals of child-bearing age born in the United States know their SCT status. "First, parents are routinely notified by NBS (newborn screening) programs if their child has SCD, but only 37 percent are notified if their child has SCT. In addition, for the parents who do receive SCT screening results, it is unknown whether they understand the implications or remember to share them with the affected child during adolescence to inform future reproductive

decisions," explained corresponding author Barry Zuckerman, MD, a pediatrician at BMC and a professor of pediatrics at BUSM. In addition, while NBS programs notify listed primary care physicians at the time of birth, results often are not readily available during routine clinic visits, and counseling or referrals to genetic counsellors are not provided in any standard fashion. Furthermore, patients may not have the same physician between birth and adolescence. The lack of knowledge of SCT status information represents a missed opportunity to provide appropriate health and prenatal counseling and testing.

"While the screening indications and contexts for life limiting diseases such as cancer, Down syndrome and SCD differ, timely knowledge of genetic vulnerability and genetic counseling are necessary for informed decision making in all screening contexts. In the case of SCD, similar to thalassemia and Tay-Sachs disease, it is important to increase the number of adolescents and young adults who know their SCT status to decrease the number of individuals inheriting SCD," said Zuckerman.

To increase awareness of SCT status and facilitate informed decision-making about reproductive options, the researchers recommend that two areas be improved in the health care system. First, the results of positive screens for SCT by NBS programs need to be reliably communicated to primary care clinicians, recorded in the patient's medical record as part of a problem list, and shared with parents as well as with the individual. The second area for improvement is to provide effective communication and information through genetic counseling on reproductive options for those with SCT.

The researchers also stress that schools and community organizations have potentially important roles in communicating the importance of SCT status for adolescents and young adults. "By working together, the health care system, schools, and community organizations may be able to improve SCT knowledge and awareness so that informed individuals

have increased access to reproductive options," he added.

Provided by Boston University Medical Center