

New sickle cell screening program for college athletes comes with serious pitfalls, experts say

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The Johns Hopkins Children's Center top pediatrician is urging a "rethink" of a new sickle cell screening program, calling it an enlightened but somewhat rushed step toward improving the health of young people who carry the sickle cell mutation.

Beginning this fall, all Division I college athletes will undergo mandatory screening for the sickle cell trait. The program, rolled out by the National Collegiate Athletic Association (NCAA), is an attempt to prevent rare but often-lethal complications triggered by [intense exercise](#) in those who carry the genetic mutation yet don't have the disease.

Nationwide, newborns are screened for sickle cell disease, but carriers, or people with one mutant and one normal sickle cell gene, do not have symptoms of the disease and may be unaware that they are carriers.

While the program's goal is laudable, its implementation has been hasty and its consequences poorly thought out, warns Johns Hopkins Children's Center Director George Dover, M.D., in a Sept. 9 commentary for The [New England Journal of Medicine](#).

The program is expected to affect nearly 170,000 college athletes and identify anywhere between 400 to 500 new cases each year. Carriers of the sickle cell trait are asymptomatic but are at higher risk for infarction of the spleen caused by lack of [oxygen supply](#) to the organ and exercise-

induced rhabdomyolysis, a condition marked by the rapid breakdown of injured muscle followed by the release of proteins in the bloodstream that harm the kidneys and can lead to [kidney failure](#). Research has shown that the risk of sudden death during exercise is between 10 and 30 percent higher among those who have the sickle cell trait than those without it. The program stems from the 2006 death of a 19-year-old freshman who died after football practice from exercise-induced rhabdomyolysis.

Dover and co-authors Vence Bonhaj, J.D., and Lawrence Brody, Ph.D., of the National Human Genome Research Institute, call the program "an enlightened first step by the NCAA toward improving the health of student athletes," but one rife with pitfalls and raising many questions. Such questions include: "Will any positive test results be followed by a second test to eliminate false positives?" and "Who is responsible for counseling students who test positive in order to explain the difference between actual disease and carrier status and the risks associated with each?"

Dover and his co-authors say that the following stipulations should be included in the program:

- Verifying test result accuracy by follow-up testing to eliminate false positives
- Post-test counseling
- Measures to prevent discrimination based on positive test results
- Making athletic practice safer to reduce or eliminate the risk for death among carriers by instituting proper hydration and avoiding workouts during high humidity and peak heat

Students will be allowed to opt out of screening if they show proof of previous testing or sign a waiver releasing their college of any legal liability. These suggest that the program was designed primarily as a legal defense measure, but its medical, social and psychological consequences remain unaddressed, the authors say.

As the most extensive sickle cell screening program in the past 30 years, this initiative will likely pave the way for other mass screening programs among college athletes, including ones aimed at identifying the carriers of cardiac anomalies, the most common cause of sudden death in athletes.

"The precedent-setting nature of this screening program dictates that we proceed with caution because any subsequent genetic screening programs may be modeled after this prototype," says Dover, a pediatric hematologist and expert on sickle cell disease.

Some 100 million people worldwide and 2 million people in the United States are believed to be carriers of the sickle cell mutation (sickle cell trait) but do not have sickle cell anemia. Named for the unusually sickle-shaped red blood cells caused by an inherited abnormality, sickle cell anemia affects nearly 100,000 Americans, most of them African-American. In sickle cell anemia, the red blood cells become rigid, which reduces their oxygen delivery to vital organs and causes them to get stuck in the blood vessels, leading to severe pain and so-called "sickling crises," which require hospitalization.

More information: The Gregory Brothers and Sickle Cell Anemia (video) www.hopkinschildrens.org/multimedia.aspx?id=6004

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