



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children
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The Honorable Kathleen Sebelius
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Sebelius:

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) is concerned about recent recommendations from athletic associations concerning the universal screening of athletes for sickle cell trait or the carrier state for sickle cell anemia. Both the National Collegiate Athletic Association (NCAA) and National Athletes Trainers' Association (NATA) have recommended sickle cell trait screening for Division 1 athletes as their method for reducing the incidence of training-related deaths. The SACHDNC supports the implementation of universal, safe training guidelines *for all athletes*, and the education of athletic coaches and trainers to recognize signs and symptoms of heat related illness and to provide prompt medical care to athletes who become ill or injured under their supervision. However, the SACHDNC is concerned about these recommendations because of the risk of stigmatization and discrimination against athletes with sickle cell trait and because the athletic associations' request for screening does not provide adequate assurance of the privacy of genetic information nor protection from the potential discriminatory use of such information.

The SACHDNC formed a *Sickle Cell Disease Workgroup* to evaluate the issues surrounding screening for sickle cell trait. Based on the workgroup's preliminary analysis, the need to single out athletes who are carriers of sickle cell trait in the athletic setting is unwarranted. Further research is necessary to understand the suggested association of sickle cell trait and the increased risk of exercise-related sudden death. The SACHDNC thinks it is important to define the clinical guidelines for carrier screening generally, and for sickle cell disease specifically.

Sickle cell diseases occur in high frequency among people of African-American, Middle Eastern, Mediterranean, Central and South American and Asian Indian origin or descent. Screening newborns for sickle cell disease and related hemoglobinopathies has been part of state-mandated newborn screening programs in the US for several decades, and for four

years it has been universal. When properly conducted (e.g., by isoelectric focusing or high pressure liquid chromatography), testing for hemoglobinopathies also detects other hemoglobinopathies and the carrier states for these hemoglobinopathies. These results have lifelong validity and, as part of general health care, individuals should be knowledgeable about their genetic risk factors for various chronic conditions and genetic disorders. Indeed, a Healthy People 2020 objective indicates that as a Nation we should “increase the proportion of hemoglobinopathy carriers who know their own carrier status.”

Sickle cell trait is associated with normal hematological values (depending on the testing performed) and normal life span. However, and although uncommon, persons with sickle cell trait may develop specific rare symptoms that may be related to sickle cell trait. Some recent reports suggest that screening for sickle cell trait outside of the newborn period may be important for individuals who are involved in high risk or strenuous activities, including sports activities, because persons with sickle cell trait may be more vulnerable to dehydration, heat stroke and muscle breakdown when subjected to strenuous exercise under unfavorable environmental conditions. Other reports indicate that deaths of healthy young athletes during conditioning are extremely atypical and are only associated with sickle cell trait in less than half of the reported cases. In addition, exercise-induced rhabdomyolysis occurs in persons without sickle cell trait. Studies of military recruits demonstrated that the incidence of exercise induced morbidity and mortality were reduced in all recruits by avoiding dehydration and adjusting the exercise intensity and rest intervals during training. In many exercise associated deaths, the deaths are attributed to sickle cell trait simply because of the presence of sickled cells in tissues at autopsy, even though the occurrence of post-mortem sickling in individuals with sickle cell trait is well documented and is expected with death from any cause.

The SACHDNC supports the implementation of universal, safe training guidelines *for all athletes*, and the education of athletic coaches and trainers to recognize signs and symptoms of heat related illness and to provide prompt medical care to athletes who become ill or injured under their supervision. The SACHDNC does not support the screening of athletes for sickle cell trait or any other genetic condition as a pre-requisite for participation in sports activities. The commonsense guidelines recommended by the Sickle Cell Disease Association of America (SCDAA) -- building up exercise intensity gradually, responding to athletes reporting symptoms of physical distress, and avoiding overheating and dehydration – if applied universally, would make athletic training safer for all and would obviate the need to identify individuals with sickle cell trait in the athletic setting. Therefore, the need to single out athletes who are carriers of sickle cell trait in the athletic setting is unwarranted. Further research is necessary to understand the suggested association of sickle cell trait and the increased risk of exercise-related sudden death.

Therefore, SACHDNC recommends that:

- All individuals should know their medical risk for various disorders, including their carrier status for various inherited genetic conditions such as sickle cell disease.
- Genetic testing or screening should not be a pre-requisite for participation in athletic endeavors.

- Evaluation and screening for sickle cell disease and other genetic conditions should take place within the individual's medical home. That evaluation should include counseling regarding the implications of the information for the individual and assurance of the privacy of genetic information.
- As part of the individual's annual medical evaluation for participation in sports, all potential athletes should receive education on safe practices for prevention of exercise and heat related illnesses.
- The Secretary, HHS, instruct SACHDNC to work with the SCDA, relevant federal HHS agencies, athletic associations, community based and health care professional organizations to develop guidelines and educational resources about screening for sickle cell trait in all persons, including athletes.
- The National Institutes of Health and the Centers for Disease Control and Prevention conduct research to ascertain if some athletes with sickle cell trait are at increased risk of exercise-related sudden death.

Sincerely yours,

R. Rodney Howell, M.D.
Chairperson