Gabriel Washington
BioC 118Q – Genomics and Medicine
Douglas Brutlag
March 8, 2011

Running the Risk: Sickle Cell Trait in Athletics

About Sickle Cell Anemia and Sickle Cell Trait

Sickle cell anemia is a homozygous dominant genetic disease that is most prevalent in populations with ancestors from malarial regions (i.e. Africa, the Mediterranean). It is "characterized by chronic hemolytic anemia, episodic painful crises, and pathologic involvement of many organs" (MeSH). Individuals with Sickle cell anemia have inherited two copies of the allele for Hemoglobin S. Hemoglobin S has a tendency to make the red blood cells form a sickle shape that can cause circulatory problems (should a sickled cell cause a jam in a blood vessel). Hemoglobin S can be particularly problematic in high stress, low oxygen conditions where more severe sickling is likely to occur. Individuals with sickle cell anemia are often advised to take precaution at high altitudes and to avoid physical exertion whenever possible.

Today, sickle cell trait, which is "the condition of being heterozygous for hemoglobin S," (MeSH) is often thought of as a benign condition. Both the name and definition of sickle cell trait imply little more than the potential to pass on the genetic allele for Hemoglobin S. Unlike the definition of sickle cell anemia, the definition of sickle cell trait does not state that the condition has any consequences of its own. Sickle cell trait understandably takes the back seat as a genetic indication of one's propensity to have a child with sickle cell anemia, which is unequivocally more serious from day-to-day than having a child with sickle cell trait. About 8% of the population in the United States of America- at least 1 out of every 12 African Americans- have the sickle cell trait. But in an increasingly homogenous society, sickle cell trait is becoming more a question of inheritance than that of race (NCAA, 2010).

Our Understanding of Sickle Cell Trait

Although it has long been known that those with sickle cell trait also have red blood cells with the potential to sickle, sickle cell trait is often referred to throughout medical literature as an innocuous condition. The severity of Sickle cell anemia compared to the relative normality of living with sickle cell trait are likely to blame for the way sickle cell trait has been neglected in medicine. The popular

understanding of the implications of sickle cell trait are demonstrated through the actions (or lack thereof) of medical professionals in dealing with the parents of the children they discover have sickle cell trait. They often fail to ensure that the parents realize that their child's sickle cell trait status is something that the child should know – this is probably because many medical professionals themselves are not aware that sickle cell trait is something that the individual needs to be aware of.

> "Perhaps trickier is whether parents know when to share the information [about their child having sickle cell trait]. Oklahoma, a leader in college-athlete testing, has detected sickle trait in 19 football players in about a decade, and only two already knew it, Anderson says. Yet when the others called home with the news, only one had a parent who hadn't known."
>
> (MSNBC, 2009)

Since 2006, all 50 states have been required to conduct tests at birth to determine a newborn's status for sickle cell anemia and sickle cell trait among other diseases (ASCAA, 2010). Although people are usually knowledgeable as to whether or not their child has Sickle cell anemia, very few are aware (or feel it necessary to inform their child) of whether they carry the Sickle cell trait or not; this is because procedure to inform parent's of their child's results is inconsistent and parents are only notified 37% of the time (Einsel, 2010). People are also unaware of their sickle cell status because Sickle cell trait is used more as a marker to assess one's potential to pass on Sickle cell anemia to their child (NCAA, 2010). Sickle cell trait is not seen as a condition with health risks of its own and carriers of Sickle cell trait are rarely thought to be at any greater risk for health complications than those with normal hemoglobin. Those with Sickle cell trait rarely receive medical consultations as to the steps they should follow to procure a brighter future for themselves. Are there any significant medical implications for Sickle cell trait? Should we be more efficient informing and consulting those with Sickle cell trait?

As of the last few years, more and more studies are demonstrating that Sickle cell trait is not as benign as was once thought. A small number of medical conditions have been linked to sickle cell trait. Furthermore, and more importantly for the topic of this paper, we have seen the dangers of sickle cell trait associated with athletic activity. With the increase of information, we have seen progress being made toward informing people of the dangers of sickle cell trait and the importance of knowing ones' status. Fortunately, sickle cell trait is slowly becoming a sign of the risk it poses to the individual and not just the risk of the individual's progeny having sickle cell anemia.

Complications with Sickle Cell Trait in Athletics

The idea that Sickle Cell Trait may present health complications arose largely as a result of a number of athlete deaths. Though infrequent, there have been enough deaths of athletes on the collegiate and even high school levels to merit interest in the subject of Sickle Cell Trait. The first known sickle cell trait-related death since the NCAA was founded in 1906 occurred in 1974. A football player from the University of Colorado, named Polie Poitier died as a result of sickle cell trait complications after the intense physical exertion of a timed mile run. There have been 21 deaths of collegiate football athletes since 2000. Nearly %50 (atleast 9) of the 21 deaths were clearly associated with sickle cell trait (Einsel, 2010). These deaths are not limited to football. In total, there have been about 15 cases of sickle cell related sudden death in the last 3 or 4 decades. These cases include track, basketball and football athletes alike (NCAA, 2010). Despite these deaths, Instances of sickle cell trait-related death seem to be increasing with time.

Biological Mechanisms

The mechanisms of health complications and the sudden death of athletes with sickle cell trait are not completely understood, but there are a few proposed mechanisms for how these issues may occur. In almost every instance, athletes that experience health complications or that collapse because of sickling have engaged in maximal exertion for at least a few minutes with little to no resting intervals (Eichner, 2010). Athletes will usually experience unusual cramps, pain or weakness and may sometimes stop before finishing the workout. Stopping because of these warning signs may help to save their lives. Those who push on despite the biological warning signs run the risk of collapsing.

There are a few nonfatal medical conditions associated with sickle cell trait that demonstrate that sickle cell trait is not completely innocuous. In fact, “for every fatal sickling collapse [in football], we hear of maybe three to five nonfatal collapses” (Eichner, 2010). Sickle cell trait-related complications include:

- One such complication is splenic infarction which is the death of tissue in the spleen due to blockage in blood flow (NIH).
- Leg compartment syndrome, (which has also been reported as problem during military training for those with sickle cell trait) is a painful condition that occurs when pressure within the muscles builds to dangerous levels. Said pressure can decrease blood flow and prevent oxygen from reaching nerve and muscle cells (NIH).

- Gross Hematuria, or visible blood in the urine. (NCAA, 2009).

If an athlete continues to push themselves despite warning signals from their body, exertional rhabdomyolysis may occur. In exertional rhabdomyolysis, red blood cells begin to take on a sickle shape that can then logjam in the blood vessels. The logjam prevents blood from flowing to the tissues normally and so the tissues become oxygen-deprived. This results in acidosis because of the buildup of lactic acid. The red blood cells may also become dehydrated. All of this can cause a buildup of lactic acid which can result in rapid muscle breakdown, called rhabdomyolysis. As the muscle tissue is degraded it can cause the onset of hyperkalemia (too much potassium) which can result in renal failure, arrhythmia and can even cause one's heart to stop.

Precautions in Athletics

The NCAA has recently taken commendable measures when it comes to the safety of collegiate athletes. Below are some of the most recent, proactive decisions:

- **June 2009**: The NCAA recommends "that member institutions test student-athletes to confirm their Sickle Cell Trait status if that information is not already known."
- **April 2010**: An NCAA mandate for the test is passed, with an amendment allowing athletes to decline by signing a waiver.
- **August 2010:** The requirement for universal NCAA screening takes effect." (Einsel, 2010)

Along with recommending that sports programs confirm the Sickle Cell Trait status of their athletes, the NCAA has generated a list of precautionary measures for athletes with Sickle Cell Trait. Following these guidelines, athletes with sickle cell trait can reach optimum performance while maintaining a healthy life. The guidelines are as follows:

The Student-Athlete with Sickle Cell Trait

- Use adequate rest and recovery between repetitions, especially during "gassers" and intense station or "mat" drills.
- Not be urged to perform all-out exertion of any kind beyond two to three minutes without a breather.
- Be excused from performance tests such as serial sprints or timed mile runs, especially if these are not normal sport activities.
- Stop activity immediately upon struggling or experiencing symptoms such as muscle pain, abnormal weakness, undue fatigue or breathlessness.
- Stay well hydrated at all times, especially in hot and humid conditions.
- Maintain proper asthma management.
- Refrain from extreme exercise during acute illness, if feeling ill, or while experiencing a fever.
- Access supplemental oxygen at altitude as needed.
- Seek prompt medical care when experiencing unusual distress.

The NCAA is in the process of developing an educational video for the membership and its student-athletes on the topic of sickle cell trait and athletics.

From the *NCAA Sports Medicine Handbook 2009*

The NCAA's suggestion that all students be offered "counseling on the implications of sickle cell trait" is particularly admirable (NCAA, 2009). Whereas the trait has often been disregarded, the NCAA is progressing toward recognizing all of the implications of the condition. This sort of activism can only benefit the health of athletes with sickle cell trait.

Precautions in Medicine

Compared to the NCAA, the health care industry has not seemed to make much progress in the treatment of sickle cell trait. Back in 1987 The National Institute of Health recommended that all newborns be screened for hemoglobinopathies like sickle cell anemia and trait. It was not until 2006 that all states became required to screen for sickle cell at birth (Einsel 2010). Furthermore, the process by which hospitals deliver this information to parents needs work. As mentioned earlier, a study by the American Journal of Medical Genetics showed that parents were only notified 37% of the time.

Moving Forward

As we progress in health care policy, it is vital that a few changes be made for the benefit of those with sickle cell trait and their families. It is time that this condition not be overlooked as completely benign, particularly when it comes to athletics. Genetic consultations, as recommended by the NCAA are a necessary step to improving the lives of those with sickle cell trait. In order for genetic

consultations to be effective, the infant screening system needs to be improved so that people are informed that their child has sickle cell trait and what exactly that may imply for their child. It can be as low as $5 to find out one's status for sickle cell trait. Seeing as 8% of the population has the sickle cell trait and many of that 8% are athletes, it is important that we make the expenditures necessary to find and educate those who may be at risk. Maybe we can even change the definition of sickle cell trait to include the complications that it has the potential to present during rigorous physical activity.

Works Cited

American Sickle Cell Anemia Association. (2010). *Newborn Screening*. Retrieved March 4, 2011, from ASCAA: http://www.ascaa.org/newborn_screening.asp

Associated Press. (2009, August 3). *More Health News*. Retrieved March 7, 2011, from MSNBC: http://www.msnbc.msn.com/id/32274232/ns/health-health_care/

Eichner, E. R. (2010). Sickle Cell Trait in Sports. *Current Sports Medicine Reports* , 347-351.

Einsel, D. (2010, July 2). Lawsuit prompts NCAA to screen athletes for sickle cell. *USA Today* .

MeSH. (1991). *NLM Controlled Vocabulary*. Retrieved March 3, 2011, from NCBI: http://www.ncbi.nlm.nih.gov/mesh?term=sickle%20cell%20trait

NCAA. (2009). *NCAA Sports Medicine Handbook.* Indianapolis: The National Collegiate Athletic Association.

NCAA (Director). (2010). *The Student-Athlete with Sickle Cell Trait* [Motion Picture].

NIH. (n.d.). *Compartment Syndrome*. Retrieved 2011, from Medline Plus.

NIH. (n.d.). *Splenic Infarction*. Retrieved 8 2011, March, from Medline Plus: http://www.nlm.nih.gov/medlineplus/ency/article/001293.htm