Awareness & Action: Integrating Genetic Testing for Sickle Cell Trait in Community Health Center, Inc.

Meriden, CT (Census Tracts 1701 & 1702)

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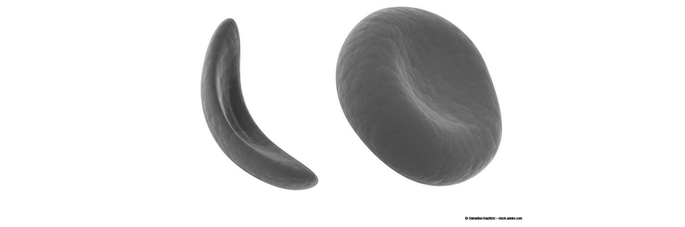
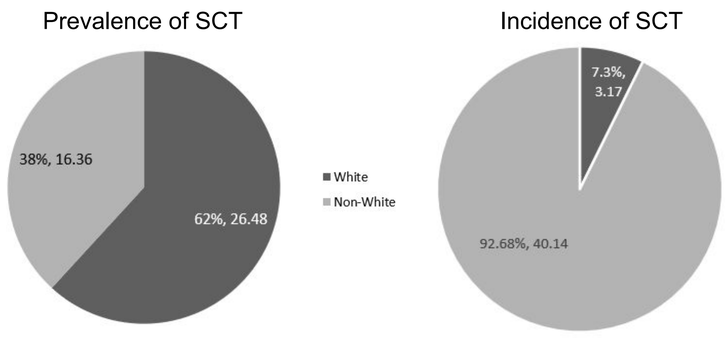
Background

Sickle cell trait (SCT) is defined as having one abnormal hemoglobin beta gene allele whereas **sickle cell disease (SCD)** affects individuals with two mutated copies of the hemoglobin beta alleles (1). It is identified by detecting the presence of **HbS (sickled hemoglobin)** which results from a switch from glutamic acid to valine, an amino acid that makes red blood cells more prone to sickle at low oxygen levels, thereby causing **organ ischemia** upon repetitive attacks (1). The determinants of SCT are largely genetic since this is a **hereditary disease**; an individual with SCT inherits hemoglobin S gene from one parent and a normal hemoglobin A gene from the other (1). This makes individuals with SCT a carrier since they have one “copy” of the mutated gene and can **pass it on to their children**, who can either have SCT or SCD (depending on the allele inherited from the spouse). Patients with SCT are often asymptomatic and typically do not present with “sickle cell/vaso-occlusive crises" unless in states of acute distress or malignancy (1). As a result, they often have a better quality of life and no increased risk of mortality compared to the general population (1).



Burden Summary



- Non-white populations have a greater incidence and prevalence of SCT & SCD than their white counterparts (1, 2)
- Census tracts 1701 and 1702 have a greater proportion of non-white residents than Connecticut on average and thus a greater burden of SCT and SCD warranting intervention
- Low-income communities where socioeconomic, health maintenance, and health literacy factors limit one’s ability to eat healthy and exercise can increase sequelae of SCT & SCD (1)

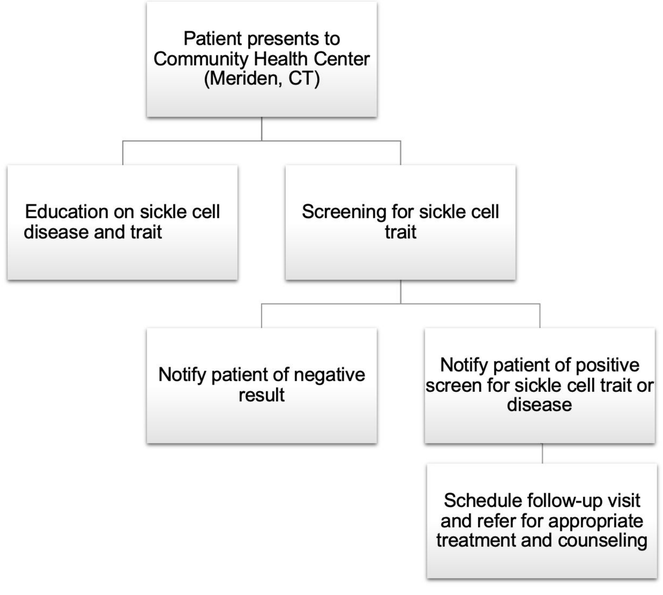
Evidence-Based Intervention

Screening for SCT has been initiated within many populations and has been successful in identifying the inherited condition prior to any associated adverse events in various settings.

Since August of 2010, the NCAA has recommended that *all NCAA athletes are screened for the condition before starting their training*. Identification of SCT is not only an important piece of public health information, but knowledge of the condition could be used to identify signs and symptoms of physical distress. It has been found that the risk of exertional death was 15 times higher in SCT+ athletes when compared to SCT- athletes (3).

SCT screening for military service has been a controversial topic within the military population. Supporters highlight that screening programs can identify individuals who may be predisposed to exertional rhabdomyolysis or sudden exertional death. Evidence supports a 40-fold increased risk of sudden death among military trainees with SCT during basic training. Opponents of SCT screening programs highlight tangible financial costs that do not outweigh possible benefits of identification and potential harms of genetic discrimination both on an individual and institutional level (4).

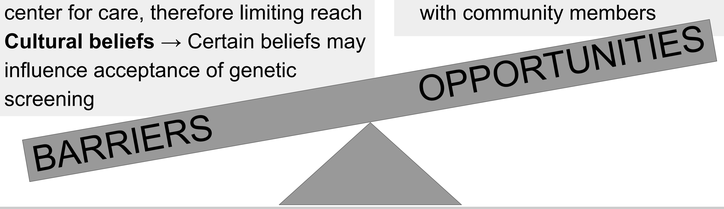
SCT screening programs have been successful in various settings, including healthcare (5, 6) and therefore should be implemented to increase awareness of SCT status within the community served by CHC, Inc. in Meriden, CT. Awareness of SCT status will not only increase overall awareness of genetic status, but also connect affected individuals with appropriate individuals to maximize current and future lifestyle choices. A flow-chart of the proposed program is as follows:



Implementation

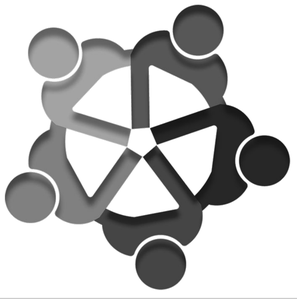
- **Costs** → Costs of testing, follow-up counseling and potential medical interventions could pose a barrier to implementation of a SCT screening program
- **Stigma** → Patients may be reluctant to participate for fear of discrimination based on testing results both socially and medically
- **Access** → Individuals with SCT may not present at the community health center for care, therefore limiting reach
- **Cultural beliefs** → Certain beliefs may influence acceptance of genetic screening

- **Integration into routine care** → efficient and cost-effective use of health care services
- **Increased awareness about SCT and benefits of screening** → can dispel myths and improve access to care
- **Reproductive counseling** can be provided for affected individuals
- **Community engagement** → opportunity to reduce stigma associated with SCT and build trust with community members



Engaging Stakeholders

- **Preventive healthcare services** →multidisciplinary team of healthcare professionals (physicians, nurses, social workers, and community health workers)
- **Community education programs** →better understanding of the disease, nutrition, diet, and exercise
- **Genetic counseling** → informed decisions, carrier status, and family planning



Policy & Practice Implications

- **Informed consent** → individuals must be fully informed, decisionally capable to make a specific and voluntary decision to participate in the SCT program
- **Ethical considerations** → privacy, confidentiality, and stigmatization concerns must be addressed and prevented
- **Follow-up counseling** → specific procedures for follow-up counseling need to be in place to ensure proper support after diagnosis
- **Community engagement** → stakeholders must engage with local community members in a culturally competent manner to address concerns, provide education, and ensure program acceptance

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