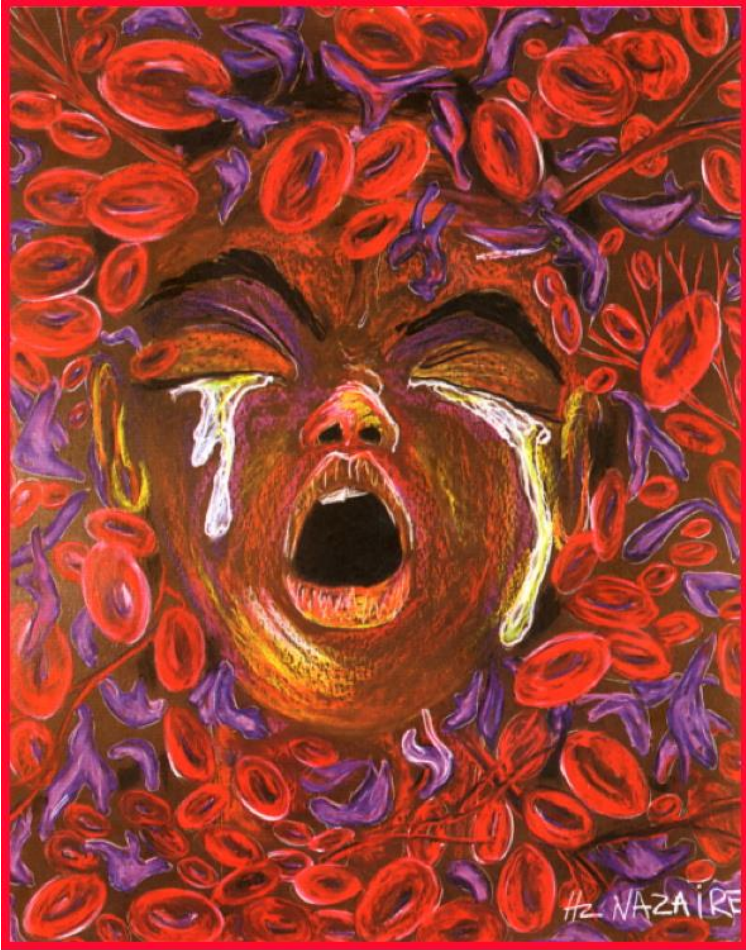


SICKLE CELL TRAIT



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SICKLE CELL TRAIT PREVENTS MALARIA, A LEADING CAUSE OF GLOBAL MORTALITY

- Sickle cell trait NEVER becomes sickle cell disease



King Tut, first case of sickle cell disease

SICKLE CELL TRAIT: A PROFESSOR AND OLYMPIAN'S FAMILY STORY



Dr. Kwaku Frempong, his wife Janet, and son

Dr. Frempong is a world-renowned expert in sickle cell disease and President of the Sickle Cell Foundation in Ghana -- and an olympic champion in track.

In 1972 --before sickle cell education and screening programs were initiated – he was diagnosed with sickle cell trait after the birth of his son, who had sickle cell anemia.

THE HISTORY OF SICKLE CELL TRAIT AND DISEASE

- **1700**: West African tribes describe sickle disease.
- **1910**: Walter Noel, a Grenadian dentist, has sickle cells in blood—coining the disease “sickle cell anemia”.
- **1923**: Sickle cell is a genetic disorder.
- **1933**: Tuskegee experiment.
- **1942**: Sickle cell trait is not a disease, but a carrier condition and is not the same as sickle cell disease.
- **1970**: Dr. Scott: “Sickle cell is high prevalence, low-priority.”
- **1972**: Charles Whitten organizes the SCDA and a movement of community sickle cell trait counselors.
- **1973**: The civil rights movement and Black Caucus initiates and demands the sickle cell anemia bill and national screening programs begin.

SICKLE CELL TRAIT

- Sickle cell trait is an inherited mutation that protects against malaria.
- It is not a disease and cannot become sickle cell disease.
- 3 million Americans are carriers: 10% African-Americans, 1% of Hispanics.
- Since 1988 in California, ~200,000 newborns with sickle cell trait, and ~9,000 with sickle cell disease
- There are 200-300 million carriers worldwide.
- Sickle cell trait individuals live normal lifespans, **generally** without problems related to sickle cell trait.
- However, here are some medical complications that *if you are knowledgeable about your trait status*, can be prevented or minimized.
- Almost all newborns in the United States undergo testing for sickle cell disease and sickle cell trait. However, many healthy adults who don't know their sickle cell trait status may have a newborn with sickle cell disease.
- Understanding one's sickle cell trait status is essential to good health.

ADEQUATE COUNSELING FOR SICKLE CELL TRAIT IS A PUBLIC HEALTH PROBLEM

- Pre-conception and prenatal counseling should be offered to all pregnancies.
- Understanding potential health issues (dehydration with severe exercise, high altitudes, hyphema, and hematuria).
- Education concerning athletic pursuits

SIGNS AND SYMPTOMS OF SICKLE CELL TRAIT

- Most people with sickle cell trait have no clinical or routine laboratory findings and do not require any change in their lifestyle or employment options
- Those with sickle cell trait have normal lifespans with no increased rate of hypertension, diabetes, or heart failure. They can donate blood and be a transplant donor.

Potential Health Issues:

- Under extreme conditions of very high altitude, deep sea diving, severe dehydration, and/or very intense physical activity in an unconditioned person, significant sickling can occur.
- Rarely, increased pressure in the eyes (glaucoma) may follow eye injuries (hyphema).
- Hematuria (blood in urine).
- Hyposthenuria (decreased ability to concentrate the urine).
- Sickle cell trait can falsely lower hemoglobin A1C in people who have diabetes but it does not cause or worsen diabetes.
- A very rare, unusual type of kidney cancer called renal medullary carcinoma.
- Not proven, but potential small increased risk of pulmonary embolism and maternal pre-eclampsia.

SICKLE CELL TRAIT AND ATHLETIC PARTICIPATION

- Those with sickle cell trait can live active lifestyles and many are professional athletes and Olympic runners.
- There have been rare individual cases of sudden death in athletes with sickle cell trait. In general, these cases occurred with severe exertion and were associated with dehydration and lack of conditioning. Some sports agencies responded by requiring sickle cell trait testing for athletes – particularly in college.
- In contrast, medical professional societies and large epidemiology publications did not recommend sickle cell trait testing.
- Preventative interventions for all athletes include: drinking adequate fluids, taking breaks when needed and a gradual adaptation to marked altitude change were recommended and demonstrated to be universally successful in military training programs, without any special attention or identification of sickle cell trait.

PUBLIC HEALTH ISSUES

- Newborn screening programs and health institutions largely do not have adequate counseling for sickle cell trait individuals -- in part because of lack of public advocacy and community demand. This results in under-education of the community about sickle cell trait and its preventable health issues.
- Sickle cell trait counseling services have dramatically decreased in the State of California. 10 regional counselors have been replaced by one telephone counseling service.
- Physicians and health providers are inadequately trained in counseling people with sickle cell trait.
- Nationwide only 16% of at-risk people know their sickle cell trait status and only 37% of families received sickle cell trait status information.
- In a survey of pediatric practices, 28% of primary care doctors did not seek or follow up on the sickle cell trait newborn status.
- Large percentages of primary care providers have not been adequately trained in counseling sickle cell trait individuals and many physicians prefer patients be referred to genetic counselors.