



Diagnosis of sickle cell trait

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SICKLE CELL TRAIT

DIAGNOSIS

Sickle cell trait is a benign carrier condition, usually with none of the symptoms of sickle cell anemia or other sickle cell diseases. However, knowledge of sickle cell trait is important in many settings, such as preconception counseling and evaluation of rare complications.

Genetics

Sickle cell trait is a generally asymptomatic carrier condition in which one allele of the beta globin gene (HBB) carries the sickle hemoglobin mutation and the other allele is normal, producing hemoglobin AS (Hb AS). If the other beta globin allele carries the sickle mutation or another pathogenic variant in HBB such as Hb C or a beta thalassemia variant), the individual will have a sickle cell disease (sickle cell anemia, sickle-beta thalassemia).

Prevalence

Prevalence of sickle cell trait in various regions around the world includes the following:

- African Americans – 7 to 10 percent
- Sub-Saharan Africa – approximately 30 percent
- Latinos – 0.2 to 6.3 percent
- India – up to 13 percent
- Middle East – 0.2 to 27 percent
- Greece – 1.5 to 7.5 percent
- Caribbean countries – 4 to 10 percent

Clinical consequences

- Sickle cell trait does not cause bone or joint pain.
- It can be associated the following findings:
- Cardiovascular disease or risk factors

- Hypertension
- Diabetes
- Heart disease
- Stroke
- Urologic and kidney disease
- CKD and albuminuria
- Hyposthenuria
- Hematuria
- UTI
- Renal medullary carcinoma
- Hyposthenuria can increase the risk of dehydration and nocturnal enuresis.
- Renal medullary carcinoma is a rare and aggressive form of non-clear cell renal cancer seen almost exclusively in individuals with sickle cell trait.
- Vaso-occlusive phenomena
 - Splenic infarction
 - Priapism
 - Complications of traumatic hyphema

Vaso-occlusive pain is not a feature of sickle cell trait. Individuals with vaso-occlusive pain should be evaluated for the possibility of sickle cell disease,

Findings not associated with sickle cell trait

Individuals with sickle cell trait do not have an increased risk of:

Vaso-occlusive pain

Sudden death

Overall mortality

References

1.E Vichinsky. Sickle cell trait. Uptodate