VERTEX

Disease Education: Sickle Cell Disease and Beta Thalassemia

SCD advocate Mapillar Dahn with her three daughters who live with SCD

What are beta thalassemia and sickle cell disease?

The inherited blood disorders beta thalassemia and sickle cell disease (SCD) result from mutations in the beta-globin *(HBB)* gene, which encodes a key component of hemoglobin, the oxygen-carrying molecule in blood.

Both diseases require lifetime monitoring and intervention as determined by medical teams. They can result in debilitating symptoms and reduced life expectancy.

Blood disorders caused by mutations in the beta-globin gene

Thalassemic | Normal | Sickled

High morbidity and mortality

Anemia | Pain | Organ Damage | Reduced Life Expectancy

Beta thalassemia

Beta thalassemia is a genetic disorder caused by a mutation in the beta-globin *(HBB)* gene. This mutation results in low or no beta-globin production, an important building block of hemoglobin, which leads to anemia.

Sickle cell disease

Sickle cell disease (SCD) is a genetic disorder caused by a mutation in the *HBB* gene that leads to an abnormal hemoglobin, called sickle hemoglobin (HbS). Because of this abnormal hemoglobin, red blood cells can become rigid and block small blood vessels.

Beta thalassemia: The patient experience

Beta thalassemia is a serious, life-threatening inherited genetic disease that is thought to affect around 288,000 people worldwide. Transfusion-dependent beta thalassemia (TDT) is estimated to affect 1 in 100,000 individuals in the U.S. general population. People with beta thalassemia are often diagnosed before age 2, based on characteristic symptoms and a set of tests, including blood tests and genetic testing. Transfusion-dependent beta thalassemia is the most severe form of beta thalassemia and requires frequent, lifelong blood transfusions. Due to anemia, people living with TDT may experience fatigue and shortness of breath, and infants may develop failure to thrive, jaundice and feeding problems. Complications of TDT can also include an enlarged spleen, liver and/or heart, misshapen bones and delayed puberty. Treatment for beta thalassemia is personalized and depends on the severity of disease that each person experiences. Many people have to get regular blood transfusions to deliver healthy donated blood to their body. This requires many long hospital visits and can also lead to an unhealthy buildup of iron. Iron overload can cause several long-term complications and requires additional treatment such as iron chelation therapy.

The lifetime costs of living with beta thalassemia

In the U.S., the lifetime costs for those living with TDT ranges from $5-$5.7 million with an average number of transfusions per year ranging from 12 to 20. The majority of these costs are for disease management such as iron chelation therapy and blood transfusions.

In addition to this financial burden, living with TDT also has a significant impact on health-related quality of life and other daily experiences. People with TDT report feeling “hooked” to the health care system due to the burden of managing their disease, impacting their ability to travel and spend time with family and friends, and committing a median of 15.8 hours per month managing their condition. They report missing 2.1 days of work per month or 2.8 school days per month due to their disease. Approximately 30% of people with TDT have been unemployed or unable to work due to their condition.

“We schedule everything around thalassemia - at night we have to make sure there’s 12 hours to take the chelator. I spend a lot of time scheduling transfusions for all of us and they’re hard to move.”

- Tracy,

Living with TDT and mother to three daughters with TDT

"Unfortunately, that's the kind of care that so many Sickle Cell Warriors are faced with, not being believed that they're in pain, not being believed that they have sickle cell. []...they often think that we are drug seeking [...]."

- Kevin,

Living with SCD, describing a stroke experience

Sickle cell disease: The patient experience

Sickle cell disease (SCD) is a serious, life-threatening, and progressive inherited genetic disease, with a global incidence of 300,000 to 400,000 infants with SCD born each year. There are approximately 100,000 people in the U.S. thought to have SCD. In the United States and Europe, most newborns are screened for SCD at birth, while symptoms usually present shortly after birth. SCD causes severe pain, organ damage and shortened life span due to the misshapen or "sickled" blood cells. People with SCD experience several symptoms in addition to pain crises, including strokes, anemia, jaundice and symptoms of heart failure. People with SCD often have spleen damage, which puts them at risk for bacterial infections. Most often, treatment is focused on relieving pain, hydration, managing fevers/infections and minimizing organ damage, requiring medication and sometimes monthly blood transfusions and frequent hospital visits.

The lifetime costs of living with SCD

In the U.S., costs of living with SCD over the lifetime are estimated to be $5.2 million, with the majority of these costs attributed to the care associated with acute complications (vaso-occlusive crises (VOCs), acute chest syndrome and stroke). However the lifetime costs of managing SCD are highly correlated with the the number of VOCs a person experiences; for example with a range of 5 to 9 VOCs per year, the lifetime costs range from $4.2 to $6.2 million, and those with significantly elevated number of VOCs (14) are thought to have $8.6 million in lifetime direct healthcare costs.

In addition to this financial burden, living with SCD also has a significant impact on health-related quality of life and other daily experiences. Patients with SCD report 27% lower average self-reported health relative to the U.S. general population, and experience severe fatigue, with fatigue scores similar to those reported by patients with anemia and cancer. 61% of people with SCD have been unemployed or unable to work due to their condition, and in the last seven days report missing an average of 12.6 hours of work due to SCD. Further, approximately 68% of people living with SCD report being treated unfairly while seeking care due to their race or need for more pain medicine.