Rare patients with sickle cell disease live nearly twice national average

October 4 2016

With a national median life expectancy of 42-47 years, people with sickle cell disease (SCD) face many challenges, including severe pain episodes, stroke, and organ damage. Compounding these complications is that SCD—an inherited, lifelong blood disorder characterized by rigid and sickle-shaped red blood cells that stick to the blood vessels, blocking blood flow—has only one FDA-approved treatment, no widely available cure, and some people may have limited access to appropriate care.

However, a new report published online today in *Blood*, the Journal of the American Society of Hematology (ASH), shows that some people with mildly symptomatic SCD may live long lives with proper management of the disease, including strong family support and strict adherence to medication and appointments. This analysis of four case studies details the outcomes of four women with milder forms of SCD who have far surpassed the U.S. median of 47 years old for women with the disease, instead living as long as 86 years.

Report author Samir K. Ballas, MD, Professor Emeritus in the Department of Medicine at Sidney Kimmel Medical College at Thomas Jefferson University in Philadelphia, hopes that this example can serve as a blueprint for others living with SCD. "For those with mild forms of SCD, these women show that lifestyle modifications may improve disease outcomes," said Dr. Ballas. Of the women described in this report, three were treated at the Sickle Cell Center of Thomas Jefferson University, and one in Brazil's Instituto de Hematologia Arthur de Siqueira Cavalcanti in Rio de Janeiro. Though they had dissimilar

ancestries (two African American, one Italian American, and one African Brazilian), all led healthy lives bolstered by long-term family support, to which Dr. Ballas attributes their long lives and high quality of life.

"It is very likely that their healthy lifestyles were important contributors to their longevity. All of the women were non-smokers who consumed little to no alcohol and maintained a normal body mass index. This was coupled with a strong compliance to their treatment regimens and excellent family support at home," said Dr. Ballas.

For this report, treatment compliance was based on observations by health care providers, including study authors. Family support was defined as having a spouse or child who provided attentive, ongoing care.

Another common factor among these four women is that they had what Dr. Ballas called "desirable" disease states. "These women never had a stroke, never had recurrent acute chest syndrome, had a relatively high fetal hemoglobin count [which helps to prevent cells from sickling], and had infrequent painful crises. Patients like this usually—but not always—experience relatively mild SCD, and they live longer with better quality of life."

As they had relatively mild disease states, none of the women were qualified to receive treatment with hydroxyurea (HU), the only FDA-approved treatment for adults with SCD. Accordingly, these patients received standard treatment including hydration, vaccination (including annual flu shots), and blood transfusion and analgesics as needed. Patients were encouraged to attend regular follow-up visits, not to smoke, watch their weight, and maintain a support system as needed.

Dr. Ballas was quick to point out, however, that this does not mean these

women lived crisis-free lives. Each experienced disease-related complications necessitating medical attention, like occasional acute chest syndrome, a problem that can cause fever, cough, excruciating pain, and shortness of breath.

It is worth noting, said Dr. Ballas, that the report does have limitations. For one, there were only four participants, all of whom were women. "Adult females with SCD generally live longer than males, but we do not know why. One possibility is that women tend to have relatively lower blood viscosity due to their lower hemoglobin level compared to males." That said, Dr. Ballas was quick to point out that this does not mean men cannot benefit from a healthy lifestyle.

To that end, Dr. Ballas hopes that patients who have defied expectations like these four women can serve as positive examples for SCD patients of all ages: "I would often come out to the waiting room and find these ladies talking with other SCD patients, and I could tell that they gave others hope, that just because they have SCD does not mean that they are doomed to die by their 40s—that if they take care of themselves, and live closely with those who can help keep them well, that there is hope for them to lead long, full lives."

Provided by American Society of Hematology