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## Genetic Services Policy Project

### Sickle Cell Disease: A Vignette

Pearl Jones is a 35-year-old African American woman with sickle cell disease (SCD). No one else in her family has the disease. Pearl was diagnosed with “sickle cell anemia” when she was 3 years old after being hospitalized for a serious infection. At the time, the doctors gave her parents a dire prognosis, informing them that Pearl was unlikely to live past 18 years of age. Pearl’s diagnosis came in the mid-1970s, when the government initiated numerous publicly-funded screening programs for sickle cell. At the time, confusion about SCD and sickle cell trait led to discrimination in employment and other settings. Pearl’s parents didn’t tell many people about Pearl’s illness because they were concerned about her father losing his job if people knew about sickle cell in their family. And although Pearl’s father was employed, finances were tight, which forced the family to rely on public assistance for Pearl’s care.

As a child, Pearl had frequent hospitalizations for pain crises. She also had an episode of acute chest syndrome, a serious lung complication. Fortunately, she did not experience any strokes or other neurological complications. Growing up, Pearl missed many days of school because of her illness, but eventually earned her GED and then an associate’s degree in office administration.

Pearl and her family lived in an area where she had access to one of the ten federally funded comprehensive sickle cell centers. In this setting, she received cutting edge treatment by sickle cell specialists, treatment that was not as readily available in other community settings. In 1992, when she was 20 and having frequent pain crises, one of her physicians at the sickle cell center suggested that she enroll in a drug study. At first, her mother and grandmother discouraged her from participating given the negative history of medical experiments involving African Americans (e.g., Tuskegee syphilis studies). Pearl, on the other hand, trusted her doctors and decided to enroll. The two-year study evaluated the efficacy of hydroxyurea, a drug previously used in cancer treatment. Pearl, who was randomized into the treatment arm of the study, had a good response to the medication with significant reduction in her pain episodes. Since then, she has remained on the medication, except during her pregnancy. The medication requires that she see her hematologist on a regular basis to monitor her blood counts, since the drug may impair production of blood cells. She continues to have periodic but infrequent episodes of pain, particularly in association with stress. Several times, the pain has been so severe she has gone to the emergency room (ER) for care.

Pain has been a challenge for Pearl as it is for many individuals with SCD. The intensity of symptoms, as well as the uncertainty about when the pain will come, often takes a significant toll. Emergency treatment of pain is frequently inadequate, with many providers underestimating the need for pain medications. Pearl believes that pain played a major role in the dissolution of her marriage several years ago. She feels that her husband never really understood the disease and wasn’t able to support her during painful episodes. Her pregnancy with her now 10-year-old son was especially difficult, and left the couple emotionally drained. Pearl turned to her mother and sisters for help caring for her young son.

Several years ago, Pearl was invited to work as an office assistant for the local sickle cell association in her community. The job has grown to involve serving as an SCD patient and family advocate as well as raising funds and organizing educational programs about the disease. Despite decades of government-sponsored programs, newborn screening, and prenatal services, Pearl notes that awareness of the disease is still limited. She sees a history of disparities in funding and political support, especially in relation to other conditions such as cystic fibrosis.

As an advocate, Pearl monitors online message boards and SCD forums, and has learned about the issues that others with the disease are dealing with. Financial concerns are a frequent topic, particularly for adults. Many adults have ongoing or increasing health problems from disease complications, but they don't always qualify for public insurance programs or disability benefits. Even if they do qualify for Medicaid, it can be difficult to find a physician who will accept the low reimbursement rates along with the challenge of managing a complex chronic disease. Access to physicians and other health care providers with experience taking care of adults with SCD is also limited. Pearl's experience in the emergency room is echoed by others she communicates with online. Employment issues come up frequently as well. The disease can make it challenging to complete one's education and obtain and maintain a job with good benefits. Despite some legal protections, there is still concern about potential employment discrimination or being fired for missing work because of the illness. The message boards are not all about complaining though. Pearl is interested in recent postings about a new nutritional supplement that seems to have helped some people. She checked out the website for the product, which says the supplement was genetically engineered to work specifically in African Americans with SCD. She is a little skeptical, but who knows...this could be the wonder drug.

## Case Issues for Discussion

1. Sickle cell disease (SCD) is often used to highlight concerns about racial discrimination in health care, employment, etc.

- a. From Pearl's experience, what evidence is presented to support these concerns?
- b. How could public policy address these concerns?

2. This scenario reflects the perspective of a patient and advocate who has had access to good care through a comprehensive SCD center.

- a. How might the story differ if told by an individual with SCD who did not have access to a comprehensive care center?
- b. How might the story differ if told by a health care provider in an area with few SCD patients?
- c. How might the story differ if told by an individual with SCD if the individual relies on state assistance versus not relying on assistance?

3. Pearl finds that adults with SCD often have difficulty accessing quality services.

- a. What factors contribute to this situation?
- b. Are the issues facing adults with SCD different than issues facing other adults with chronic health conditions? If so, how are they different?
- c. What health care provider educational deficits are highlighted by this scenario?
- d. What services and supports would assure that adults with SCD receive appropriate care?

4. Pearl had frequent absences from school as a child because of her SCD.

- a. What other social ramifications are associated with this disease?
- b. Are there adequate supports to address these concerns? If not, what additional resources would be beneficial?
- c. Do you think the stigma Pearl and her family experienced in the 1970s still exists today? Please explain.

5. At the end of this scenario, Pearl hears about a new nutritional supplement that has been getting press on the online message boards frequented by people with SCD. She does additional research and finds the website for the product and seems convinced that this could be helpful to her.

- a. What are the potential benefits and harms of online message boards and other Internet resources as educational and informational sources?
- b. What issues or concerns does the nutritional supplement raise?
- c. Should such supplements be regulated?
- d. Should advertising for supplements be regulated?

What genetic service delivery issues does this scenario raise?

- Historical issues with screening programs and research projects
- Comprehensive care centers vs. community treatment (geographic disparities in care)
- Pediatric to adult transition issues

What provider issues are identified?

- Provider knowledge of disease (e.g., inadequate pain management in the ER)

What consumer issues are identified?

- Consumer knowledge base, availability and usefulness of information
- Support for social consequences of disease (e.g., divorce support groups, educational information for families)
- Concerns about racism, discriminatory treatment in health care
- Confusion regarding trait vs. disease

What payer or coverage issues are identified?

- Reliance on public assistance programs (SSI)

What industry issues are identified?

- Development of targeted therapies (“soft science” issues)

What other policy issues are raised by this scenario?

- Regulation of nutraceuticals (use of “genetics” in the marketing of nutritional supplements, lack of clinical testing for supplements)
- Differential power and resource base of different groups (sickle cell vs. cystic fibrosis)
- Education and employment accommodations, alternate arrangements
- Employment discrimination