SM Journal of Hematology & Oncology

Letter to the Editor

Carriers in Hemophilia: The Invisible Link between Generations

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DEAR EDITOR,

Hemophilia X-linked bleeding disorder, caused by deficiencies in clotting factors VIII (1:5,000) and FIX (1:30,000) in males. In females, lyonization leads to random X-chromosome inactivation, which increases the risk of bleeding associated with the factor VIII/ IX genes. The typical symptom experienced by females is heavy menstrual bleeding with other clinical manifestations of postpartum hemorrhage. However, most bleeding in females is undetected due to a lack of awareness. To determine the correct clinical features of haemophilia, the diagnosis should be accurate. Earlier screening tests to detect bleeding related to hemophilia include one-stage clotting assays, followed by molecular genetics analysis, which help to provide accurate diagnostic methods based on quantification. The genetic analysis, particularly for bleeding with mild phenotypes, was initially focused on carrier detection. The new approach, HA with female patients, involves molecular analysis that begins with X-inactivation analysis by PCR amplification, followed by direct sequencing or Sanger sequencing of the F8 coding region. Recently, next-generation sequencing (NGS) has become available for the advanced genetic diagnosis of bleeding disorders. It is capable of detecting large deletions and duplications using multiplex ligation-dependent probe amplification. Sequencing of F8 and F9 to determine carrier status and significance using advanced diagnostic methods. Genetic testing is crucial for females in this condition, as they require more broad information on family history, factor levels, and X-inactivation patterns. In the future, females will be able to overcome their underreporting symptoms and reproductive challenges during prenatal and postnatal bleeding conditions. One of the main challenges of haemophilia carriers is inhibitor development to control the bleeding episode, which becomes more complex, mainly when they receive replacement factor therapy; however, genetic diagnosis is precise about the inhibitor levels. Molecular genetic diagnosis to understand the clinical management and prognosis of the patient's condition and their family relative situation. Without analysing the genetics, it is challenging to use the test to identify the exact cause of the bleeding with a mild phenotype. Nowadays, modern diagnosis is crucial for identifying low factor levels in combination with symptoms and carriers of the gene. Genetic analysis is the gold standard for clinical management to identify carrier status in an unrevealed female carrier. To identify female carriers of hemophilia and understand their experiences and the challenges that may not be extensively recognised, it is essential to explore the situations, including the physical and mental impact of living with hemophilia. Raising awareness of the diagnosis among females significantly enhances their quality of life.

Submitted: 07 September, 2025 | **Accepted:** 17 September, 2025 | **Published:** 18 September, 2025

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Citation: Elayaperumal S, Keerthi G (2025) ACarriers in Hemophilia: The Invisible Link between Generations. SM J Hematol Oncol 7: 1.