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LETTER

Exploring Female Relatives of Patients with Hemophilia' Awareness, Attitudes, and Understanding Towards Genetic Testing [Letter]

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Dear editor

In clinical practice, hemophilia is likely to be underdiagnosed and misdiagnosed. Because of its rarity, there are insufficient data on hemophilia to assist its management.[1] The study by Zhao et al, regarding awareness, attitudes, and understanding towards genetic testing in patients with hemophilia (PWH), was read, reviewed, and much appreciated by our group.[2] The discovery of the genetic abnormalities that cause hemophilia is crucial for family counseling since it predicts bleeding propensity and inhibitor risks. Genetic testing has increased the number of families with diagnosed defects, while also improving carrier testing and prenatal diagnosis.[3] In their study, Zhao et al tried to gain a better understanding of the knowledge and attitudes of female relatives of PWH towards genetic testing, through qualitative interviewing and/or thematic analysis, which could provide information to support the design and development of genetic counseling services, as well as facilitate the implementation of genetic testing plans for female relatives of PWH. This study is really promising for healthcare systems to manage the diagnosis and treatment of PWH, even though only a small number (11) of female relatives of PWH was included in the study. In addition, this study revealed the limited background knowledge and information in the family members and relatives regarding the genetic test for hemophilia diagnosis and treatment. However, the subjects included in this study still showed positive perceptions and attitudes towards the genetic test, which may be surprising for such a medical issue.

However, the question raised here is whether this outcome could have a wider impact worldwide, not only in China; therefore, the medical efforts regarding hemophilia diagnoses and treatment should be performed specifically for those who are being targeted. The complex technical and human infrastructure required for good disease diagnosis, treatment, and monitoring is tough enough, even in prosperous countries. However, this type of high-technology medical diagnosis and precision medicine requires high-tech medical equipment and well-trained personnel, and is more difficult to perform in low- and middle-income countries.[4] Therefore, researchers in this field could consider giving information at a certain level and medical suggestions for countries with these limitations.

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Disclosure

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