EDITORIAL

Diagnosis and data under the spotlight

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In this second edition of *The Journal of Haemophilia Practice*, Robin Sager questions the diagnostic labeling of women with inherited bleeding disorders [1]. She asks whether a haemophilia carrier with symptoms is a haemophilia carrier, a symptomatic carrier or a woman with a bleeding disorder. Furthermore, she ponders whether a diagnostic label of "woman with haemophilia" or "woman with a bleeding disorder" would afford such women better access to healthcare and outcomes more like those we expect for males with haemophilia. But would such a revision risk health care services losing contact with asymptomatic carriers? And how good are we at supporting the clinical, genetic counselling and/or psychosocial needs of mothers, sisters and daughters of males with haemophilia, whether symptomatic or not?

The United Kingdom Haemophilia Centre Doctors' Organisation (UKHCDO) is currently attempting to register all known carriers with our national haemophilia database, regardless of factor level or symptoms. The strategy aims to enable a better understanding of the size of the cohort of women at risk of having a baby with haemophilia, which may result in better obstetric care for at-risk infants.

Kaspar suggests that for every person with haemophilia there are 1.6 haemophilia carriers [2] so the UK carrier population is likely to be about 10,850. Identification of actual or potential carriers per male with haemophilia requires up to date family trees and knowledge of kinship.

The policy will affect three groups of women:

- Daughters of men with haemophilia (obligate carriers)
- Daughters of carriers who have low factor levels
- Daughters of carriers who have normal levels and whose carriership would only be revealed by genetic testing.

The first two groups can be said to be carriers and to be at risk, but should we treat them all the same? Robin Sager argues that carriers with low factor levels should be acknowledged as women with haemophilia and identified differently from asymptomatic carriers.

Tracking these identified women raises logistic and personal issues. Women have to be found, contacted, invited to clinic and counselled about testing and the value of data collection. Clearly, some will not wish to know they have an underlying genetic condition: knowledge of a genetic condition may affect the individual's ability to obtain health or life insurance, [3] may impact on future marriage prospects and reproductive choices, or (for those past child-bearing age) may be considered irrelevant.

Should newborn daughters of men with haemophilia be registered at birth? Clearly this would require a mechanism for removal of the individual from the register in the event that paternity is not confirmed, or consent is withdrawn. The issue of how and when to broach consent from minors are challenging. Genetic testing advice in the UK recommends testing of at-risk individuals be delayed until they are "Gillick competent", and fully understand the implications of testing and consent to it, as well as to being registered as an affected individual (if applicable) [4,5]. For younger, asymptomatic girls, centres assume parental consent to screen to be sufficient. But, should a parent be able to consent to revealing the genetic status of their asymptomatic daughter, thereby removing any future, potential self-determination by the daughter? Anecdotally, there is considerable national variation in screening practice in this area. By contrast, for boys with haemophilia there is no concern about genetic information being used for screening of at-risk women in his family.

Appropriate factor level measurement in an "at risk" girl requiring surgery would identify them as a carrier without genetic testing if levels were found to be low. Equally, family trees can identify obligate carriers without prior knowledge, consent or discussion. In some cases, this is beneficial [6].

Data management is fundamental to many aspects of haemophilia care. The UK national database contributes positively to national guidelines and academic work, and provides an immediate point of reference for clarification of disease status in times of clinical uncertainty. The importance of data custodianship on the ever-evolving backdrop of database security and governance policy is of paramount importance. The European Court of Justice recently ruled that individuals who object to data held on them by technology companies have "the right to be forgotten". Do our patients have the same right? Our challenge as haemophilia care providers combines the ongoing importance of identifying and engaging forgotten or overlooked female carriers while also respecting and implementing the right of those who wish to be "forgotten" again."

References

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