

Women and girls with haemophilia

Robyn Shoemark

Changing treatment, changes diagnosis

Chair ~ Kate Khair, Director of Research, Haemnet, London, UK

Recognizing women with a bleeding disorder: a new diagnosis

~ Sarah O'Brien, Assoc Prof Pediatrics, Director of Experimental Therapeutics, The Ohio State University, Nationwide Children's Hospital, Columbus, USA

Sarah O'Brien's presentation at Congress tackled some of the issues in diagnosing haemophilia in females.

She commented that haemophilia diagnosis for women has previously been ambiguous. Some practitioners have frequently been heard to say that females affected by haemophilia do not bleed and that their male relatives' bleeding disorder has nothing to do with the females' bleeding. We as bleeding disorder health care professionals know this is not true.

Girls and women affected by haemophilia need to be diagnosed correctly. If their factor level is greater than 40%, the diagnosis is *symptomatic* or *asymptomatic* carrier, depending on their bleeding phenotype (the detectable expression of the gene or their clinical symptoms). If their factor level is less than 40%, the categories of *mild*, *moderate* and *severe* that are given to males also apply to females.

In clinic, history taking is of utmost importance. There is wide variability in bleeding phenotypes amongst females. There is often no correlation between factor levels and bleeding. Females often present with heavy menses (periods), oral cavity bleeding, postpartum haemorrhage (severe vaginal bleeding after childbirth) and excessive bleeding after dental extraction. Joint bleeds and subclinical bleeds can occur. This is why clinical phenotypes are more important in females than actual factor levels. Genetic testing should be part of the

haemophilia diagnosis process for females.

The take-away from this session was the questions to use when talking with a females in clinic when abnormal bleeding is suspected. Does the female bleed more than 7 days when she menstruates, have more than 4-5 changes of sanitary products daily (make sure to clarify the products used as there is a large range of absorbency in products on the market), need to change products at night, do they have gushing when they stand up, and are there any clots and are they larger than the size of a grape? Answering yes to these questions would warrant further investigation to lead to a correct diagnosis. And don't forget that obligate haemophilia carriers may have other bleeding disorders too.



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