ABSTRACT FORM ECAT SYMPOSIUM 8 – 9 NOVEMBER 2018

Name:

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Title:

One-stage clotting assay versus chromogenic testing in the diagnosis of haemophilia patients

Abstract:

Haemophilia A and B are hereditary bleeding disorders, caused by a deficiency in factor VIII (FVIII) and factor IX (FIX), respectively. Diagnosis is primarily based on clotting factor activity assays. Most frequently used assays are the one-stage assay (OSA) and the chromogenic assay (CSA). As these assays are based on different principles and a variety of mutations are underlying haemophilia A and haemophilia B, results between assays may be discrepant within patients. Discrepancies, usually defined as a 2-fold difference, are mainly caused by specific mutations in the *F8*-gene or *F9*-gene. Both the clotting factor activity measured with the CSA or the OSA may be higher. Assay discrepancies are seen in 20-50% of haemophilia A patients and approximately in 33% of haemophilia B patients. In a group of 130 patients with HA we saw 2-fold assay discrepancies in 14.6%. CSA was higher in 18/19 discrepancies (94.7%). Discrepancies may classify patients as severe with one assay and mild with the other. Moreover, diagnosis may even be missed due to discrepancies. Unfortunately, it is not yet completely clear which assay best reflects the phenotype of hemophilia, but literature suggests to use both assay to prevent missing a diagnosis. For some mutations in hemophilia A patients, studies have shown superiority of the CSA in reflecting the bleeding phenotype, for other mutations it may be the OSA. In hemophilia B, less data are available, but the bleeding phenotype seems to correspond with different assays in different patients.

To conclude, assay discrepancies are seen frequently and may cause wrong test results or a change in treatment plan. Therefore, keeping them in mind is important, especially if the test results do not reflect the bleeding phenotype.