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Title **Value of Mutation Detection and of the Activated Protein C Resistance Assay in Inherited Thrombophilia**

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Aim

To assess the diagnostic value of the activated protein C resistance (APCr) test to advise French National Health Insurance (NHI) on reimbursement.

Conclusions and results

We selected 5 studies on the diagnostic performance of the APCr test (2155 patients), 7 guidelines on mutation testing, and 1 impact study. We found no study addressing the direct impact of testing on morbidity and mortality. Sensitivity of the APCr test was 100%, and specificity was 68% to 100%. In 3 studies, the positivity threshold was defined *post hoc* for 100% sensitivity. According to the working group, the diagnostic performance of marketed APCr tests varies. In clinical practice, mutation detection either replaces or follows the APCr test. The indications for mutation testing are: (a) in men and women under 50: unexplained or recurrent deep vein thrombosis or pulmonary embolism, (b) in pregnant women: an episode of venous thrombosis, or a personal or proven family history of venous thrombosis.

Recommendations

In the opinion of HAS (French National Authority for Health), mutation testing should replace the APCr test. Both genetic tests should be reimbursed by NHI.

Methods

We reviewed published data on (a) the diagnostic benefit of the tests (safety, efficacy, and contribution to treatment strategy), and (b) their public health benefit (impact on morbidity and mortality). The review was discussed by a working group of 8 hematologists and then submitted to the HAS Committee for Assessment of Medical and Surgical Procedures for their opinion.

Further research/reviews required

The following are required:

- additional data on benefits and risks
- a list of the APC resistance tests with good diagnostic performance.

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