

An ACCE review: Genetic testing for Long QT syndrome

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Background: Policy decisions regarding the provision of a genetic test should be informed by a comprehensive evaluation of this test. A formal structure for test evaluation, known as the ACCE framework, was developed by the Centre of Disease Control and Prevention (USA). This framework is made up of 44 questions including information on the disorder and setting, analytical validity, clinical validity, clinical utility and ethical, legal and social issues.

Aim: To evaluate genetic testing for Long QT Syndrome in Australia using the ACCE framework. This will be used to inform policy regarding the provision of genetic testing for Long QT in Western Australia.

Method: The published literature and some grey literature on Long QT was reviewed in order to answer the 44 questions posed by the ACCE framework. The review was also informed by local expertise.

Outcomes: Analytic sensitivity and specificity were found to be 75% and 100% respectively. The test needs to be validated in the Western Australian population, and negative and positive predictive values determined for this setting. The value in screening for LQTS lies in the identification and treatment of asymptomatic family members prior to coronary event. Guidelines are available for the management of LQTS.

Conclusion: Although the comprehensiveness of the ACCE review was restricted by the evidence available, the review was considered to provide sufficient evidence for genetic testing for Long QT. As the service develops in Western Australia and more evidence becomes available through this and internationally, the ACCE review can be developed further.