Ontario
Health Quality Ontario
Qualité des services de santé Ontario

OHTAC Recommendation: Genetic Testing for Predisposition to Dilated Cardiomyopathy

Ontario Health Technology Advisory Committee

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Issue Background

Dilated Cardiomyopathy

Dilated cardiomyopathy (DCM) is a condition in which the heart becomes weakened and enlarged, affecting its ability to pump blood efficiently. At least 1 in 2,500 individuals currently live with DCM; however, the condition is considered largely underdiagnosed since subjects often remain asymptomatic until serious symptoms arise. Both in children and in adults, DCM is a common cause of heart failure. In children, DCM is the most common type of heart muscle disease.

Genetic Testing for Dilated Cardiomyopathy

Genetic testing for dilated cardiomyopathy is a complex, multistep process. Testing is typically performed to identify whether an individual with DCM harbours a specific gene mutation that may be causing their disease. Testing is therefore meant to confirm whether the cause of an individual's DCM is genetic, but it also helps to identify whether that individual harbours mutations that also raise his/her risk of arrhythmia or syndrome features. If a genetic mutation that causes DCM is identified, family members of the individual may be screened for the same mutation using a more targeted, single-gene genetic test. Family members with a disease-causing mutation who do not currently show symptoms of the disease may be at risk of developing DCM later in life. Identification of these high-risk family members could allow for heightened clinical surveillance and potentially, early intervention. However, the true impact of testing on clinical outcomes and management has not been established and requires further research. As of the date of this guidance, 33 genes have been identified as being associated with DCM. The complicated genetic nature of DCM has implications on the sensitivity (also known as the mutation detection rate) of genetic tests. The sensitivity of a genetic test for DCM depends on the number of genes being sequenced as well as the sequencing methodology. Next generation sequencing methodologies for this indication are currently emerging and require further research.

Summary of OHTAC Findings

No trials met the inclusion/exclusion criteria for the primary questions of the Medical Advisory Secretariat's evidentiary review. A full evidence-based review was therefore not completed. However, preliminary evidence projects a low test sensitivity (of less than 30%). The clinical test sensitivity as well as the clinical utility of genetic testing (i.e., whether testing improves health outcomes or alters disease management) therefore remain uncertain. Further research is needed.

OHTAC Recommendations

In considering the lack of evidence at this time regarding the diagnostic accuracy and clinical utility of genetic testing for DCM (i.e., the impact of testing on clinical outcomes and disease management both for index cases and/or their family), OHTAC makes the following recommendation:

- OHTAC does not recommend access to genetic testing for DCM for individuals diagnosed with this disease or for their immediate or extended family members.