



Repetitive Constitutional Genetic Testing

SYNOPSIS AND RELEVANCE

Constitutional genetic testing (CGT) should only be performed once in a patient's lifetime except in very unusual circumstances. Efforts to avoid repeat CGT can:

1. Prevent performing repetitive CGT when such testing provides no added value to the patient.
2. Impact the costs for CGT to medical facilities, medical practices, and patients.

INSIGHTS

1. Constitutional genetic tests are usually performed only once because an individual's genetic composition does not change within their lifetime.
2. Repeating a constitutional genetic test may be considered in the following circumstances:
 - a. The first genetic test result must be confirmed because it did not match the clinical picture (ie, phenotype/genotype mismatch).
 - b. In patients who have undergone stem cell or solid organ transplant when the donor tissue is thought to alter the 'constitutional' genetics of the patient (eg, stem cell transplant in patients with von Willebrand disease).
 - c. The previously performed test did not fully explain the patient's phenotype and was less comprehensive than the current test, which incorporates new variants and/or genes. In these cases, it may not be possible to test only for the new gene/variant targets.

BACKGROUND

The American College of Medical Genetics and Genomics, as part of the Choosing Wisely initiative of the American Board of Internal Medicine, recommends against duplicate constitutional genetic testing (CGT) for inherited conditions unless there is uncertainty about the validity of the existing test result or some rare clinical situations. Although the human genome is stable, repetitive CGT commonly occurs and adds little value to caring for patients with genetic disorders.

We now better understand why tests are unnecessarily repeated. Before electronic health records (EHR) were widely implemented, clinicians often repeated genetic and other laboratory tests because they did not know the test was already performed, or did not have access to the test results. Duplicate constitutional genetic testing may also occur when these tests are included in order panels that are built to ease ordering for healthcare providers. In these instances, the tests in the order panel that are medically-indicated at that time may obscure the genetic test that was already performed. The test will be repeated unless the ordering provider knows to remove the genetic test from that order. Of course, there are rare medical indications for repeating a constitutional genetic test, and ordering systems should allow for such circumstances.

The increasing use of the EHR to order and result laboratory tests affords the opportunity to determine the extent of repetitive genetic testing within an institution. Moreover, clinical decision support tools that exist may be adapted to reduce the ordering of repetitive genetic testing.

REFERENCES

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