An Evidence Framework for Genetic Testing (National Academies Report)

Recommends a framework for decision-making regarding the use of genetic tests in clinical care.

Updated last November 20, 2017
for the 03/27/2017 report.

WHAT IT DOES

In light of the rapid development of genetic testing and its use in medical practice, the National Academies of Sciences, Engineering, and Medicine (NASEM) convened a committee at the request of the Department of Defense (DOD)’s Office of Health Affairs. The committee was tasked with evaluating the evidence base for genetic testing and providing recommendations on how to make decisions regarding use of genetic testing for clinical care. They published these evaluations and recommendations in a report entitled “An Evidence Framework for Genetic Testing.”

NASEM outlined two sets of recommendations. First, they suggested that the DOD support internal and external research to improve the understanding of the validity and usefulness of present and future genetic tests. The second set of recommendations is a framework for making decisions about the use of genetic tests for clinical care:

1. Define the “genetic test scenario” (the type of test, the purpose, the population, etc.);
2. Prioritize the “genetic test scenarios”:
   - Is the test worthwhile?
   - Does the available evidence support its use?
   - What are the contextual issues (e.g., social, ethical, legal)?
3. Review the evidence;
4. Decide if the test is appropriate for coverage;
5. Assemble a repository of decisions to review previous decisions and to evaluate new scenarios more efficiently;
6. Periodically review and revise previous decisions; and
7. Identify evidence gaps and consider how to fill them.

The DOD Office of Health Affairs can use these recommendations to improve the Military Health System, which serves 9.6 million military members, veterans, and their families.

RELEVANT SCIENCE

Genes are units of information, made of DNA, that provide instructions for the development and function of living organisms. They allow for characteristics to be passed on from one generation to the next. Mutations are changes that alter the DNA sequence, and they accumulate in a person’s DNA over the course of that person’s lifetime. Most of these changes have no effect, however sometimes mutations can impact health and development.

According to the NIH, genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. Genetic tests can be used for various purposes:

1. Diagnostic: to determine if an individual has a particular genetic condition (e.g., a sequence-based test can confirm a diagnosis for retinitis pigmentosa);
2. Predictive: to identify risk of developing inherited disorders (e.g., women with certain BRCA1 variants are at increased risk for developing breast cancer); and
3. Reproductive: to identify risk of an individual passing on a genetic disease to their child, or to identify an embryo or fetus with a genetic condition (e.g., a genetic test to confirm suspicions of a trisomy disorder, such as Down syndrome, in a developing fetus).

In order to evaluate newly developed genetic tests, the NASEM identified the measures that need to be considered:

- Analytic validity: does the test measure the genetic information that it claims to measure?
- Clinical validity: does the test accurately and reliably predict the clinical presentation of interest?
- Clinical utility: does the test improve clinical outcomes or provide other value to the patient?

WHY IT MATTERS

The result of genetic testing can inform individualized prevention and treatment strategies. However, the rapid development of new tests poses various challenges, including the validation of their use in clinical settings. In addition, the NASEM report indicated concerns over:

- Costs: How do we balance the cost of genetic tests with their benefit? Will use of genetic tests increase health care expenditure?
- Potential to increase inequality in healthcare: Are current and near future genetic tests more clinically useful for people of European decent, on whom most of the research has been conducted?
- Ethical, legal, and social implications: Is there an obligation to inform family members of genetic risks? Will people be stigmatized or socially distanced as a result of genetic testing?

RELEVANT EXPERTS

Sharon F. Terry, MS, is President and CEO of Genetic Alliance.

Relevant publications:


BACKGROUND

The Human Genome Project spelled out nearly the entire sequence of the human genome for the first time in 2003. Since then, various stakeholders have developed a large number of genetic tests to identify genetic variants associated with disease. Over 54,000 tests for more than 11,000 conditions have been reported to The National Institutes of Health (NIH) Genetic Testing Registry (GTR).

The NASEM committee considered many previous frameworks when developing their suggestions for the DOD:

- The US Preventative Services Task Force method;
- The Fryback-Thornbury hierarchic model of efficacy;
- The analytical validity, clinical validity, clinical utility, and associated ethical, legal, and social implications model;
• The Evaluation of Genomic Application in Practice and Prevention framework;
• The Genetic Testing Evidence Tracking Tool;
• The McMaster University evaluation framework; and
• The Frueh and Quinn framework.

ENDORSEMENTS & OPPOSITION

Endorsement:

• Sharon F. Terry, President and Chief Executive Officer of Genetic Alliance, perspective, July 1, 2017: “I welcome active participation by the DoD in the quest to aggregate evidence that will help to determine the appropriate use of genetic tests. Essentially a large insurer, TRICARE, could provide state-of-the-art care for its members, and at the same time contribute to large databases such as ClinVar that would advance public understanding of the association between variants and phenotypes. This would advance the quality of care that the nation’s military and their families would receive, as well as provide benefit for all of the stakeholders seeking to make evidence-based decisions.”

Opposition:

• At present, there has not been any publicly reported opposition to this report.

STATUS

NASEM published this report in March 2017.

RELATED POLICIES

The Genetic Information Nondiscrimination Act (GINA) prohibits discrimination by employers and health insurance providers on the basis of genetic information.

ORGANIZATIONS

The Department of Defense’s Office of Health Affairs asked the National Academies of Sciences, Engineering, and Medicine to examine the relevant medical and scientific literature to determine the evidence base for different types of genetic tests (e.g., predictive, diagnostic, and prognostic) for patient management. The committee is to provide recommendations to advance the development of an adequate evidence base for genetic tests to improve patient care and treatment. Additionally, the committee will recommend a framework to DOD for decision making regarding the use of genetic tests in clinical care.

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