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Association for Molecular Pathology Position Statement: Direct Access Genetic Testing (Direct to Consumer Genetic Testing) - February 2015

Background:

In 2007, the Association for Molecular Pathology (AMP) published a position statement on Direct Access Genetic Testing (Direct to Consumer Genetic Testing) which concluded that genetic testing should be available only through appropriately qualified health professionals that order tests from laboratories that are certified by CLIA for high complexity testing. In 2014, the Department of Health and Human Services (HHS) finalized a new rule that gives patients direct access to test results, including genetic test results, directly from the laboratory providing the services. In addition, expanded use of health information technology with patient portals simplifies independent patient access to their health information. Genetic tests have become increasingly available for direct purchase by consumers without a professional intermediary, are marketed directly to consumers, with results sent directly to the consumer by the purveyor of the test.

These paradigm shifts, which are intended to give the general public a stronger role in preventive decisions and healthcare management, appear to be a permanent sector of the healthcare environment. Direct access genetic testing has been much debated. The information provided has the potential to motivate patients to make changes that could prevent disease, and seek professional care when a test indicates the presence of illness. However, certain testing may be merely a "hook" to sell products and services of dubious benefit. Some may have the potential to cause harm if patients or their physicians take action based on inaccurate results,

During several months in 2014, a Working Group of AMP leaders examined the current environment of direct access genetic testing and proposed that clinically meaningful tests could benefit the public, under certain conditions. Therefore, AMP has revised its position statement on Direct Access Genetic Testing.

AMP has identified four categories of direct access genetic testing:

1. Clinically meaningful: The tests provide information that can diagnose, predict, prognosticate, and/or otherwise reveal information relevant to a patient's health. [Support, under certain conditions]
2. Business interest: The information garnered from the testing does not meet criteria for 'clinically meaningful' and companies attempt to sell products or services owned and/or endorsed by the laboratory. These test providers have a secondary financial gain through purchase of supplements, books, etc. [Oppose]
3. Ancestry: Tests reveal information about biological relationships among individuals and families, but do not provide health information. [Neutral]
4. Recreational/novelty: These tests do not provide health-related information, but rather attempt to convey non-medical information, such as genetically-based differences in taste of foods. [Neutral]

AMP Supports:

Direct access to clinically meaningful genetic testing may add value to patients and consumers, when certain standards are met. Accordingly, AMP now supports direct access genetic testing for clinically meaningful tests under the following conditions:

- The association between the genetic marker for which testing is performed and the relevant disease must be robust and supported by strong scientific evidence in the peer reviewed literature, and/or be based on evidence referenced or annotated in current genetic/genomic databases.
- Testing should comply with the CLIA statute and regulations, and all applicable state and federal laws and regulations.
- Transparency regarding the analytical and clinical validity of the tests should be present in all marketing materials and included in the report. Specifications include but are not limited to, analytical and clinical sensitivity/ specificity, and the limitations of assay. These should be described in terms understandable to an educated lay reader (see next bullet point). In addition, the underlying data and analytical methodology, including computational and/or statistical methods employed, power analyses, confidence analyses, etc. upon request should be readily available to health professionals.
- Reporting of test results and the limitations of the test should be in lay language. Additionally, the report should include an interpretation of the finding and describe its significance for the consumer's health status.
- Test validation and interpretation should be performed by certified molecular laboratory professionals.
- AMP strongly supports referral for genetic counseling services and the provision of educational materials. Test providers should encourage genetic counseling as an additional step for consumer education. Resources such as referrals to the National Society of Genetic Counselors' directory or through genetic counseling contracting services are considered appropriate.
- Direct access laboratories should recommend that consumers discuss their test results with their physicians.

AMP Remains Neutral:

Recreational/novelty and ancestry testing create educational opportunities for the public to learn and expand its understanding of genetics and inheritance. Because the information garnered from these tests typically does not include health information, AMP has chosen to remain neutral on consumer direct access to these types of tests.

AMP Opposes:

Direct access genetic testing that provides information that is not clinically meaningful **and** that businesses use to sell additional products and services. AMP opposes the direct to consumer marketing of these types of tests. AMP believes that consumers and healthcare professionals should be discouraged from ordering these types of tests and recommends that policy mechanisms be created to ensure that marketing materials and reports clearly identify the lack of data to support health management and the secondary business interests associated with the test.