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# Association for Molecular Pathology Position Statement: Consumer Genomic Testing – June 2019

***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 for healthcare decision making should be available only through appropriately qualified health professionals that order tests from laboratories that are certified by the Clinical Laboratory Improvement Amendments (CLIA) for high complexity testing. In 2015, AMP revised the position statement to support direct access genetic testing, provided certain conditions are met.

Since that time, the consumer genomics market has continued to expand and evolve, which warrants AMP to further update its position statement. A working group of AMP leaders evaluated the current landscape of consumer genomic testing. The results of that assessment reveal updates and expansions to the conditions that should be met by consumer genomic testing providers offering and delivering clinically meaningful genomic testing results directly to consumers. The phrase 'consumer genomic testing' used in the rest of this statement is intended to mean any test that is marketed and delivered directly to the consumer, which may or may not be ordered by a physician.

As in 2015, AMP identified four categories of consumer genomic 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: 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:***

AMP supports consumer genomic testing for clinically-meaningful tests, which may benefit consumers under the following conditions:

- All health-related claims must have well-established clinical validity. The gene-disease association is robust and supported by strong scientific evidence in the peer reviewed literature, and/or is based on

evidence referenced or annotated in current genetic/genomic databases (e.g., ClinGen criteria[1,2] for definitive or strong gene-disease association).

- The laboratory must comply with the CLIA statute and regulations, including having documented analytical validity, a robust quality management system, and appropriately licensed or credentialed laboratory personnel. Test validation and interpretation should be performed by board-certified molecular laboratory professionals.
    - The underlying data and analytical methodology, including computational and/or statistical methods employed, power analyses, confidence analyses, etc. should be readily available to health professionals upon request.
- Transparency regarding the analytical and clinical validity of the tests should be present in all marketing materials and included in each consumer's report of laboratory results.
    - Specifications include but are not limited to, analytical and clinical sensitivity/specificity, and the limitations of assay such as residual risk of disease in the absence of genetic variations.
- Reporting of test results and the limitations of the test should be in lay language. As an example, different technologies used by consumer genomic companies—single-nucleotide polymorphism (SNP) array, targeted genotyping, next generation sequencing have different limitations, failure modes and breadths of coverage, which should be described so consumers and their health care providers can determine the limitations of the test.
- The report should include an interpretation of the finding(s) and describe its significance for the consumer's health status, as well as implications for family members.
- Additionally, some negative test findings only reduce disease risk for well-defined ethnic subgroups or patients and are not applicable to the broader population, if applicable this information should be explained to the consumer as well.
- AMP recommends that consumer genomic testing providers adhere to The Future of Privacy Forum's "Privacy Best Practices for Consumer Genetic Testing Services."[3]
- Consumer genomic testing providers are encouraged, but not required, to conduct evaluations to gauge user comprehension of test description materials and test results, including ethnic-specific residual risk interpretations.
- AMP strongly supports referral for genetic counseling services and the provision of educational materials to consumers of genomic testing. Test providers should provide genetic counseling or encourage genetic counseling as an additional step for education of the consumer (and the consumer's family, if applicable). Genetic counseling resources such as referrals to the National Society of Genetic Counselors' directory or through genetic counseling contracting services are considered appropriate.
- Consumer genomic testing providers should recommend that consumers discuss any actionable test results with their physicians for clinical care decisions. Positive potentially-actionable test results should be discussed with a healthcare provider, who may order confirmatory or additional testing prior to initiating treatment.
- Consumer genomic testing providers should provide clear language about potential insurance risks, e.g. the Genetic Information Nondiscrimination Act (GINA) protects against health insurance discrimination, but not long-term disability or life insurance.

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[1] https://www.clinicalgenome.org

[2] Strande *et al.* (2017) Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence –Based Framework Developed by the Clinical Genome Resource. American Journal of Human Genetics. 100(6):895-906.

[3] The Future of Privacy Forum, Privacy Best Practices for Consumer Genetic Testing Services, 2018: https://fpf.org/2018/07/31/privacy-best-practices-for-consumer-genetic-testing-services/

***AMP Remains Neutral:***
Recreational, novelty, lifestyle, and ancestry testing may 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 these types of consumer genomic tests.

***AMP Opposes:***
Consumer genomic testing that provides information that is not clinically valid and/or that businesses use to sell additional products and services, such as unsubstantiated claims concerning athleticism, diet, exercise, or cosmetics. AMP opposes marketing of these types of consumer genomic 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.