Consumer-Initiated Use of Genomic Services

Proposal for Short-Term Action

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Proposed Action Step

- Develop a brief document that reviews main concerns about direct-to-consumer (DTC) testing, such as
  - Limited data on clinical validity and utility of tests
  - Consumer and provider understanding of test results
  - Privacy protections
  - Companies that skirt oversight regulations
  - False and misleading claims
- Highlight recommendations from SACGHS reports that address concerns

Relevant SACGHS Recommendations

7 recommendations from the oversight and coverage and reimbursement reports address major concerns:

- Limited clinical validity and utility data: 2 recommendations
- Consumer and provider education: 3 recommendations
- Privacy protections and companies that skirt regulations: 1 recommendation
- False and misleading claims: 1 recommendation

Clinical Validity Recommendation

- The Committee is concerned by the gap in oversight related to clinical validity and believes that it is imperative to close this gap as expeditiously as possible. To this end, the Committee makes the following recommendations:
  - FDA should address all laboratory tests in a manner that takes advantage of its current experience in evaluating laboratory tests.
  - This step by FDA will require the commitment of significance resources to optimize the time and cost of review without compromising the quality of assessment.

Clinical Validity Recommendation (continued)

- The Committee recommends that HHS convene a multistakeholder public and private sector group to determine the criteria for risk stratification and a process for systematically applying these criteria. This group should consider new and existing regulatory models and data sources (e.g., New York State Department of Health Clinical Laboratory Evaluation Program). The multistakeholder group should also explicitly address and eliminate duplicative oversight procedures.
- To expedite and facilitate the review process, the Committee recommends the establishment of a mandatory test registry.

Clinical Utility Recommendation

- HHS should create and fund a sustainable public/private entity of stakeholders to assess the clinical utility of genetic tests (e.g., building on CDC's Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative). This entity would:
  - Identify major evidentiary needs
  - Establish evidentiary standards and level of certainty required for different situations such as coverage, reimbursement, quality improvement, and clinical management
  - Establish priorities for research and development
  - Augment existing methods for assessing clinical utility as well as analytical and clinical validity, such as those used by EGAPP and the U.S. Preventive Services Task Force, with relevant modeling tools

Clinical Utility Recommendation (continued)

- Identify sources of data and mechanisms for making them usable for research, including the use of data from electronic medical records
- Recommend additional studies to assess clinical effectiveness
- Achieve consensus on minimal evidence criteria to facilitate the conduct of focused, quick-turnaround systematic reviews
- Increase the number of systematic evidence reviews and make recommendations based on their results
- Facilitate the development and dissemination of evidence-based clinical practice guidelines and clinical decision support tools for genetic/genomic tests
- Establish priorities for implementation in routine clinical practice
- Publish the results of these assessments or otherwise make them available to the public via a designated HHS or other publicly supported Web site (e.g., GeneTests)

Clinical Utility Recommendation (continued)

- To fill gaps in the knowledge of the analytical validity, clinical validity, clinical utility, utilization, economic value, and population health impact of genetic tests, a Federal or public/private initiative should:
  - Develop and fund a research agenda to fill those gaps, including the initial development and thorough evaluation of genetic tests and the development of evidence-based clinical practice guidelines for the use of those tests
  - Disseminate these findings to the public via a designated HHS or other publicly supported Web site (e.g., GeneTests)

Education Recommendations

- HHS should work with all relevant government agencies and interested private parties to identify and address deficiencies in knowledge about appropriate genetic and genomic test applications in practice and to educate key groups such as health care practitioners, public health workers, public and private payers, and consumers of health care. These educational efforts should take into account differences in language, culture, ethnicity, and perspectives on health and disability as well as issues of medical literacy, access to electronic information sources such as the Internet, and deficiencies in public infrastructures (e.g., libraries) that can affect the use and understanding of genetic information.

Education Recommendations

- Since genetic tests and services are being integrated into all areas of health care and since providers have an important role in ensuring appropriate use of and access to genetic tests and services among diverse populations, there is a critical need for programs to educate and train health care providers and payers in genetics and genomics. Health care providers should be able to meet established genetic competencies and, thereby, integrate genetics effectively into their practices. The HHS Secretary should develop a plan for HHS agencies to work collaboratively with Federal, State, and private organizations to develop, catalog, and disseminate case studies and practice models that demonstrate the relevance of genetics and genomics.

Education Recommendations

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- The HHS Secretary should provide financial support to assess the impact of genetics education and training on health outcomes.

- The HHS Secretary should strive to incorporate genetics and genomics into relevant initiatives of HHS, including the National Health Information Infrastructure.

Education Recommendations

- For patients and consumers to evaluate health plan benefits and health care providers and to make the most appropriate decisions for themselves and their families, they need reliable and trustworthy information about family history, genetics, and genetic technologies. The HHS Secretary should ensure that educational resources are widely available through Federal Government Web sites and other appropriate public information mechanisms to inform decisions about genetic tests and services.

Recommendation to Address Oversight and Privacy

- CLIA regulations and, if necessary, CLIA's statutory authority, along with FDA's risk-based regulatory authority and regulatory processes, should be expanded to encompass the full range of health-related tests, including those offered directly to consumers. Relevant Federal agencies (e.g., CMS, CDC, FDA, and FTC) should collaborate to develop an appropriate definition of health-related tests that FDA and CMS could use as a basis for expanding their scope. Additionally, these Federal agencies, including the HHS Office for Civil Rights, along with other State agencies and consumer groups should propose strategies to protect consumers from potential harm and from unanticipated and unwanted compromises in privacy that may lead to harm. Additional oversight strategies that might be established should be balanced against the benefits that consumers may gain from wider access to genetic tests and potential cost savings.

Recommendation to Address False and Misleading Claims

- Appropriate Federal agencies, including CDC, CMS, FDA, and FTC, should strengthen monitoring and enforcement efforts against laboratories and companies that make false and misleading claims about laboratory tests, including direct-to-consumer tests.

Next Steps for Brief Report

- Establish short-term task force to assist with developing the report

- Determine whether there are issues that would not be addressed by past recommendations
  - If so, how should they be addressed?