Testimony: National Committee on Vital and Health Statistics

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June 15, 2010

RE: Genetic Information Panel

Thank you for the opportunity to comment on the issues of privacy and security of genetic health information. The American College of Medical Genetics (ACMG) represents doctoral level laboratory and clinical geneticists certified by the American Board of Medical Genetics, the 24th primary board of the American Board of Medical Specialties. ACMG has about 1500 members, most of whom are Fellows of the ACMG. We believe that genetic health information should, for the most part, be integrated with other useful health information to maximize health service delivery. We are also interested in the questions of how genetic health information should be protected in electronic medical records while still being available to health care providers when needed for care and to investigators for the improvement of genetic health care service delivery. Our comments will be focused on heritable disease traits rather than acquired genetic traits used in the characterization of cancers and leukemias.

Genetic information related to heritable disease traits includes diagnostic information, disease susceptibility risks, reproductive risks, and family information. It can be obtained in prenatal and postnatal health care settings and in the course of public health screening programs. Individuals may be symptomatic or asymptomatic. Because genetic information includes family information, additional issues of genetic information use revolve around family members who may not be the patient. Although family history is often useful in genetic assessments and necessary for risk calculations, the information about many family members provided by a patient will have been obtained without the individual consent of other family members. This has led some genetics groups to keep family data out of the central medical records and in their own "shadow" files. The use of shadow files is less common for other types of genetic information since privacy protections have been put into place.

Genetic testing has moved from targeted genetic tests to multiplex assays of many genes at once. As technology rapidly evolves to whole genome analysis at low cost over the next five years, we are faced with how best to store the information in a secure form that protects privacy but allows access to the results for interpretation as patients present for health care services. For patients presenting with symptoms, particular parts of the genome known to be associated with the features seen could be accessed. For many patients with genetic diseases, access to genetic information may be critical for emergency department care. Pharmacogenetic data may influence choice of therapeutics and for maximal utility in an emergency situation, may have to be available electronically.. There may also be times when actionable disease risks should be identified so as to allow for appropriate lifestyle or health care modifications.

Public health uses of genetic testing have grown significantly in the past several years. Newborn screening (NBS) for genetic diseases is now the highest volume area of genetic testing with over 4.1 million newborns each year screened for 28 genetic diseases. These are typically conditions for which early medical intervention and ongoing treatment are critical. Programs to ensure access to medical information including genetic information during emergency situations are being developed. There is an enormous investment being made in health information exchange in states to facilitate the transfer of information between hospitals, providers and public health programs for these highly vulnerable patients.

Further, programs have developed to allow for the collection of longitudinal health information from patients identified in NBS in order to better understand the clinical histories of the diseases. There are important differences in how public health departments use genetic information vs. its use in personal health care. In newborn screening, there are a number of conditions for which highly efficient determination of risk followed by rapid communication between public health and private providers is essential to realizing the expected outcomes following intervention. Further, under public health mandates, data considered private and to only be obtained with patient consent can also be a useful epidemiological surveillance data point.

Numerous institutions have made various levels of medical and genetic information available to patients. This information has proved valuable in emergency medical situations and access should be encouraged. More difficult has been identifying types of genetic information over which a patient can exercise control over access. Some types of health care would not require genetic information to be available while others would. Our current knowledge of genetic diseases remains somewhat limited so it is not always clear whether a particular clinical presentation might be linked to a genetic condition. A system that allows for access to particular types of clinical information as needed would be ideal but it could be difficult to determine which information is not of direct importance. That related to reproductive risks is less likely to be needed in routine health care but may be difficult to separate. It seems likely that this can only be accomplished around broad categories of genetic information.

Summary Recommendations:

- Ensure the availability of genetic health information when needed in health care service delivery
- Anticipate that complete genomic information may become readily available at costs below the current costs of s single targeted genetic tested. Unless costs drop so far as to make retesting as needed practical, information will have to be stored for later use.