ARTICLE

Outcomes of Genetics Services: Creating an Inclusive Definition and Outcomes Menu for Public Health and Clinical Genetics Services

KERRY SILVEY,* JACQUIE STOCK, LIANNE E. HASEGAWA, AND SYLVIA MANN AU

Third party payers, funding agencies, and lawmakers often require clinicians and public health agencies to justify programs and services by documenting results. This article describes two assessment tools—''Defining Genetics Services Framework'' and ''Genetics Services Outcomes Menu,'' created to assist public health professionals, clinicians, family advocates, and researchers to plan, evaluate, and demonstrate the effectiveness of genetics services. The tools were developed by a work group of the Western States Genetics Services Collaborative (WSGSC) consisting of public health genetics and newborn screening professionals, family representatives, a medical geneticist, and genetic counselors from Alaska, California, Hawaii, Idaho, Oregon, and Washington. The work group created both tools by an iterative process of combining their ideas with findings from a literature and World Wide Web review. The *Defining Genetics Services Framework* reflects the diversity of work group members. Three over-lapping areas of genetics services from public health core functions to population screening to clinical genetics services are depicted. The *Genetics Services Outcomes Menu* lists sample long-term outcomes of genetics services. Menu outcomes are classified under impact areas of Knowledge and Information; Financing; Screening and Identification; Diagnosis, Treatment, and Management; and Population Health. The WSGSC incorporated aspects of both tools into their Regional Genetics Plan.

KEY WORDS: outcome; genetics services; genetic service; public health; quality

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INTRODUCTION

People who use clinical genetics services want to know the services are the best available. Clinical geneticists seek to understand how the quality of their care influences their patients' health. Public health professionals working in state genetics and newborn screening programs are responsible for assuring the core public health functions and essential services they perform reflect best practice. Third party payers, funding agencies, and lawmakers often require clinicians and public health genetics and newborn screening programs to justify programs and services by documenting outcomes; yet, developing a practical strategy for assessing quality and demonstrating beneficial outcomes of genetics services is challenging because genetic conditions affect people of any age and any body system. Last year a group from the United Kingdom: Katherine Payne, Stuart Nicholls, Marion McAllister, Rhona MacLeod, Dian Donnai, and Linda Davies, published a review of validated outcomes measures of clinical genetics services. They concluded that "... a core set of outcome measures may be necessary when evaluating genetics services" [Payne et al., 2008]. In a subsequent publication, the

developing a practical strategy for assessing quality and demonstrating beneficial outcomes of genetics services is challenging because genetic conditions affect people of any age and any body system.

Kerry Silvey, M.A., C.G.C., is Co-Director of the Western States Genetics Services Collaborative, and a Public Health Genetics Specialist at the Oregon Center for Children and Youth with Special Health Needs, Child Development and Rehabilitation Center, Oregon Health & Science University.

Jacquie Stock, M.P.H., is an Evaluation Consultant and Work Group Facilitator for the Western States Genetics Services Collaborative, and a Research Associate at Seattle Children's Center for Children with Special Needs.

Lianne Hasegawa, M.S., C.G.C. is the Western States Regional Genetics Services Collaborative Project Coordinator and a Genetic Counselor at the Hawaii Department of Health.

Sylvia Au, M.S., C.G.C., is Co-Director of the Western States Genetics Services Collaborative and the State Genetics Coordinator at the Hawaii Department of Health.

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*Correspondence to: Kerry Silvey, M.A., C.G.C., Child Development and Rehabilitation Center, CDRC—Clinical Services Building, 901 East 18th Avenue, Eugene, OR 97403-5254.
E-mail: ksilvey@uoregon.edu

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same group concluded that "there appears to be no agreement about appropriate outcome measures" for clinical genetics services [McAllister et al., 2008].

In 2006, participants in the Western States Genetics Services Collaborative (WSGSC) decided that improving their ability to evaluate the impact of genetics services was a high priority. The WSGSC is one of seven regional genetics services collaboratives in the United States funded by the U. S. Department of Health and Human Services, Health Resources and Services Administration. The WSGSC includes Alaska, California, Guam, Hawaii, Idaho, Oregon, and Washington. Participants include representatives from each state/territory genetics program and each newborn screening program, family representatives from each state/territory; genetics services specialists, primary care providers, and others. Several WSGSC participants have led or participated in state, regional, or national efforts to develop outcomes for children with special health care needs (CSHCN), conditions included in the newborn screening blood test, or other genetics services evaluation projects.

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The purpose of this article is to describe two tools developed by the work group—the *Defining Genetics Services Framework* (Fig. 1) and the *Genetics Services Outcomes Menu* (Fig. 2). The process used to develop the tools is also described. The tools are significant because they incorporate both public health and clinical health care delivery aspects of genetics services in the United States, and because they were created by an inclusive group of clinicians, family representatives, and public health professionals.

METHODS

The WSGSC Outcomes Work Group, comprised of Collaborative participants from Alaska, California, Hawaii, Idaho, Oregon, and Washington included one medical geneticist, two genetic counselors, three family advocates, and public health genetics, and/or newborn screening advocates from each state, as well as the WSGSC co-directors and evaluation consultants. A series of three in-person and seven "webinar" work group meetings were convened during 2006 and 2007 to develop a list of beneficial outcomes of genetics services. A broader group of WSGSC participants representing medical geneticists, genetic counselors, primary care providers, family advocates, and public health professionals contributed to the development of both tools by reviewing and commenting on drafts during discussion sessions at two annual WSGSC Regional Summit meetings and by e-mail. At its first meeting, the work group agreed upon three foundational assumptions they believed would facilitate application of the outcomes tool. The outcomes should be: (1) non-condition specific; (2) practical to measure, preferably using existing data sources, and (3) useful for clinicians, health care administrators, public health professionals, third party payers, and legislators.

Process to Develop the Defining Genetics Services Framework

During their second meeting, work group members listed and discussed genetics services and programs for which outcomes should be developed. A wide spectrum of public health activities, education efforts, and clinical services were all mentioned. The group decided the first step in developing outcomes for genetics services was to define the term

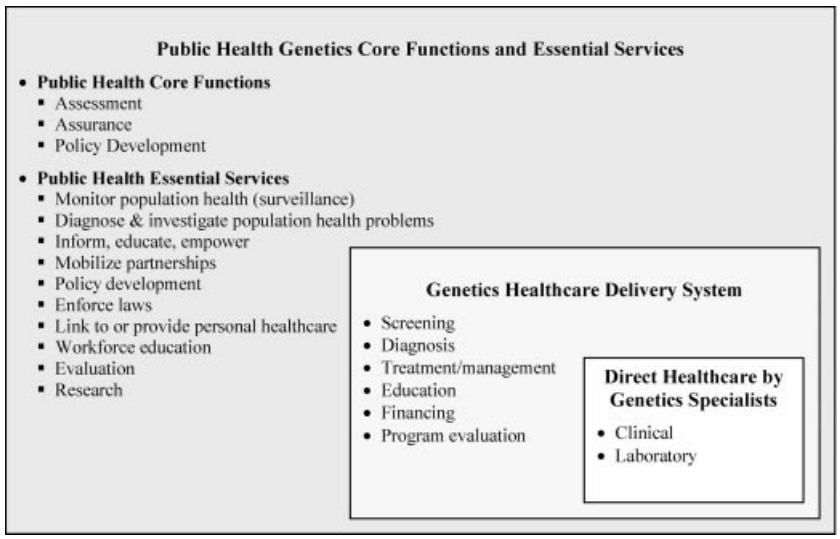


Figure 1. Defining genetics services framework. The "Genetics Healthcare Delivery System" includes "Direct Healthcare by Genetics Specialists." "Public Health Genetics Core Functions and Essential Services" encompasses both the "Genetics Healthcare Delivery System" and "Direct Healthcare by Genetics Specialists."

"genetics services." The authors were charged with conducting a peer-reviewed literature and Internet search to find published thought on the definition of genetics services. The authors prepared a draft framework of genetics services definitions based upon the work group's list of services and findings from the literature and Internet review. Work group members reviewed and refined this framework. The resulting *Defining Genetics Services Framework* was reviewed by the larger WSGSC and finalized by the authors based upon their input.

Process to Develop the Genetics Services Outcomes Menu

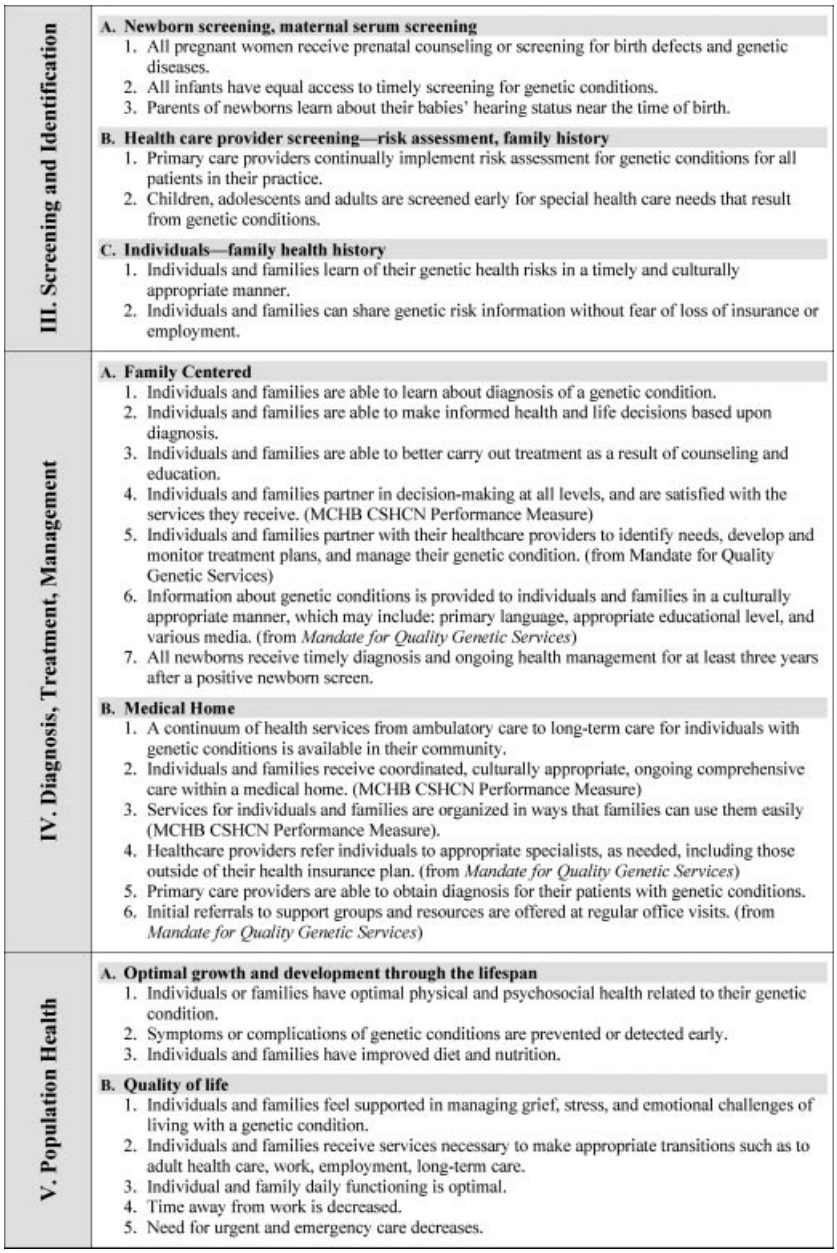
In August 2006 a review of the peer-reviewed professional literature and Internet was completed using keyword search terms "genetic services/genetics services/medical genetics/genetic counseling" and" outcomes/measurement/evaluation/ assessment." Databases used to review the peer-reviewed professional literature were MEDLINE, CINAHL, EMB Reviews, and PSYCH INFO English language articles from 1996. Based on the literature review results, project staff compiled a list of outcome concepts to present to the work group to stimulate discussion of potential outcomes. In addition, two documents—the *Mandate for Quality Genetic Services*, developed by the Genetic Alliance [Genetic Alliance, 2001], and a report describing measures of psychological and emotional benefit for patients who receive genetic counseling [Fryer and Lister Cheese, 1998] were provided in their entirety to the work group because these documents provided an overview of outcome concept areas for clinical genetics services.

The work group turned to the field of CSHCN as a model for creating outcomes for groups with diverse diagnoses [Maternal and Child Health Bureau, 1989]. The group decided to revise their original plan and develop a more comprehensive list of outcomes that users could choose from and modify based on their own needs. The work group selected outcome concepts from the results of the literature review and then brainstormed additional potential outcomes concepts they wanted to consider. The *Genetics Services Outcomes Menu* was created following an iterative process of writing by the authors, review and comment by the work group, and rewriting until consensus wording was achieved. Analogous concepts were combined into one outcome. When outcome concepts matched MCHB or *Mandate for Quality Genetic Services* performance measures, the wording of these performance measures was used. To word outcomes, the work group applied the model of "Measuring and Monitoring Community Based Systems of Care for CSHCN" [Roberts and Behl, 2001]. Work group members chose to phrase all outcomes as a standard (desired situation) rather than as a change over time.

At the 2007 WSGSC Regional Summit meeting, collaborators including family advocates, genetic counselors, medical geneticists, primary care physicians, and genetics and newborn screening public health professionals reviewed and commented upon the *Genetic Services Outcomes Menu*. At the 2008 WSGSC annual Regional Summit meeting the WSGSC Medical Home Work Group, which includes primary care physicians, physicians experienced in serving children with special health care needs, and family advocates, also reviewed the Menu. Suggestions from the Medical Home work group were incorporated into the current version of the *Genetics Services Outcomes Menu*.

Impact Area	Outcomes
I. Knowledge and Information	**A. General public** 1. Individuals and families have information about the impact of genetics on their or their family's health, and are able to make informed decisions based upon this information. 2. Quality, culturally appropriate resources exist that assist individuals and families in understanding family health history; signs or symptoms of genetic conditions; screening and testing options and implications; diagnosis; treatment; and long-term follow-up. 3. Parents are confident in communicating about familial genetic risk information with their children. 4. Individuals or families know what genetic services they need and where to find them. 5. Information about genetic research and clinical trials is available to families and integrated into clinical practice. 6. Individuals and families participate in treatment at optimal levels after receiving counseling and education. **B. Health Care Providers** 1. Health care providers use current information about incidence; prevalence; epidemiology; diagnosis; and treatment of genetic conditions to prevent, cure, and treat individuals with heritable conditions. a. Health care providers integrate information about clinical trials and research into clinical practice. b. Health care providers use up to date, diagnosis specific protocols that are available on the internet. **C. Others - public health agencies, insurers, legislators, researchers** 1. Federal and state legislators use current information about genetics to write laws that foster prevention, cure, and treatment of genetic conditions. 2. Providers, payers, and employers have policies and procedures to ensure appropriate use of genetic information. 3. Public health agencies have up to date information about incidence, prevalence and epidemiology of genetic conditions, and apply this knowledge to foster prevention, cure, and treatment.
II. Financing	**A. Insurance** 1. Financing for screening is adequate. 2. Early and timely screening contribute to lower long-term diagnostic and treatment costs. 3. Financing for diagnosis, health management and long-term care is adequate. a. Health insurers reimburse diagnostic and treatment services for genetic conditions. (from *Mandate for Quality Genetic Services*) b. Individuals and families have adequate private and/or public insurance to pay for services they need. c. Payers acknowledge psychosocial as well as physical effects of a genetic condition, on both the individual and the family, at each stage of life. (from *Mandate for Quality Genetic Services*) **B. Public health funding** 1. Funding is adequate at local and state levels to successfully carry out core public health functions and the ten essential public health services related to genetic conditions.

Figure 2. Genetics Services Outcomes Menu—Outcomes to measure the impact of genetics services.



III. Screening and Identification	**A. Newborn screening, maternal serum screening** 1. All pregnant women receive prenatal counseling or screening for birth defects and genetic diseases. 2. All infants have equal access to timely screening for genetic conditions. 3. Parents of newborns learn about their babies' hearing status near the time of birth. **B. Health care provider screening—risk assessment, family history** 1. Primary care providers continually implement risk assessment for genetic conditions for all patients in their practice. 2. Children, adolescents and adults are screened early for special health care needs that result from genetic conditions. **C. Individuals—family health history** 1. Individuals and families learn of their genetic health risks in a timely and culturally appropriate manner. 2. Individuals and families can share genetic risk information without fear of loss of insurance or employment.
IV. Diagnosis, Treatment, Management	**A. Family Centered** 1. Individuals and families are able to learn about diagnosis of a genetic condition. 2. Individuals and families are able to make informed health and life decisions based upon diagnosis. 3. Individuals and families are able to better carry out treatment as a result of counseling and education. 4. Individuals and families partner in decision-making at all levels, and are satisfied with the services they receive. (MCHB CSHCN Performance Measure) 5. Individuals and families partner with their healthcare providers to identify needs, develop and monitor treatment plans, and manage their genetic condition. (from Mandate for Quality Genetic Services) 6. Information about genetic conditions is provided to individuals and families in a culturally appropriate manner, which may include: primary language, appropriate educational level, and various media. (from *Mandate for Quality Genetic Services*) 7. All newborns receive timely diagnosis and ongoing health management for at least three years after a positive newborn screen. **B. Medical Home** 1. A continuum of health services from ambulatory care to long-term care for individuals with genetic conditions is available in their community. 2. Individuals and families receive coordinated, culturally appropriate, ongoing comprehensive care within a medical home. (MCHB CSHCN Performance Measure) 3. Services for individuals and families are organized in ways that families can use them easily (MCHB CSHCN Performance Measure). 4. Healthcare providers refer individuals to appropriate specialists, as needed, including those outside of their health insurance plan. (from *Mandate for Quality Genetic Services*) 5. Primary care providers are able to obtain diagnosis for their patients with genetic conditions. 6. Initial referrals to support groups and resources are offered at regular office visits. (from *Mandate for Quality Genetic Services*)
V. Population Health	**A. Optimal growth and development through the lifespan** 1. Individuals or families have optimal physical and psychosocial health related to their genetic condition. 2. Symptoms or complications of genetic conditions are prevented or detected early. 3. Individuals and families have improved diet and nutrition. **B. Quality of life** 1. Individuals and families feel supported in managing grief, stress, and emotional challenges of living with a genetic condition. 2. Individuals and families receive services necessary to make appropriate transitions such as to adult health care, work, employment, long-term care. 3. Individual and family daily functioning is optimal. 4. Time away from work is decreased. 5. Need for urgent and emergency care decreases.

Figure 2. (*Continued*)

RESULTS

Defining Genetics Services Framework

Although the term "genetic(s) services" appears frequently in the peer-reviewed genetics literature and in documents posted on genetics policy-related websites, the term is usually not defined. The definitions we found were based on the following criteria: (1) who provides the service; (2) who receives the service; (3) the setting where the services are provided; (4) what healthcare services are included; (5) whether only clinical services are addressed or if non-clinical public health services are included; and (6) the purpose of the definition.

Although the term "genetic(s) services" appears frequently in the peer-reviewed genetics literature and in documents posted on genetics policy-related websites, the term is usually not defined.

The *Guidelines for Clinical Genetic Services for the Public's Health* [Council of Regional Networks for Genetic Services, 1997] and the subsequent *Integrating Genetic Services into Public Health—Guidance for State and Territorial Programs from the National Newborn Screening and Genetics Resource Center (NNSGRC)* [Kaye et al., 2001] describe genetics services that should be available in a state and discusses them from multiple perspectives: family focused versus population based; clinical versus laboratory; age of individuals served (prenatal, childhood, adolescence, adulthood); and levels of prevention addressed (primary, secondary, tertiary). Both documents include services for conditions that are usually not considered genetic such as diagnosis of fetal alcohol spectrum disorder or counseling pregnant women about teratogen exposures. These services are often combined administratively in clinical or public health programs and may be provided by the same personnel as healthcare for genetic conditions.

The medical genetics services definition in the *Specialised Services National Definition Set of the United Kingdom (2nd Edition)* [Department of Health, United Kingdom, 2007] is based upon the setting in which services are delivered, that is, "In the main, this activity is provided by specialist genetic centers." Since diagnostic laboratory services, expert advice to policy makers, education of healthcare professionals, and participation in research are typical functions of genetics centers in the United Kingdom, these activities are included. Their definition specifically excludes treatment or management of metabolic conditions because metabolic biochemistry is considered a separate specialty.

The *Defining Genetics Services Framework* shown in Figure 1 is divided into

three arenas. The narrowest area of definition is Personal Healthcare Services Provided by Individuals Trained in Genetics. These services are included in the intermediate level definition, Genetics Healthcare Delivery System, which is in turn included in the broadest definition, Public Health Core Functions and Essential Services.

Public Health Genetics Core Functions and Essential Services. The broadest level of the framework is comprised of the core public health functions: assurance, assessment, and policy development as they relate to genetics; and the Ten Essential Public Health Services: monitor population health (surveillance); diagnose and investigate population health problems; enforce laws; inform, educate, empower; mobilize partnerships; policy development; link to or provide personal healthcare; workforce education; evaluation; and research [Association of State and Territorial Health Officers, 2001].

Genetics Healthcare Delivery System. This intermediate level of the framework is a combination of public health and personal healthcare activities. This level includes the public health activity of population screening as well as personal health screening by non-genetics specialists, linked with diagnosis, treatment, and management of people with or at risk for genetic conditions. Education and financing activities needed to support the population and personal health screening to medical management continuum are included in this level and provided by public health as well as private organizations and clinicians. Education activities at this level can target not only the public health workforce and the population at large, but also primary care providers, genetics specialists, policy makers, and people with or at risk for genetic conditions. The WSGSC work group drew interventions or "genetics services" provided in this second area of the framework from concepts presented in three publications: (1) *Guidelines for Clinical Genetic Services for the Public's Health* [Council of Regional Networks for Genetic Services, 1997]; (2) *Serving the Family From Birth to the Medical Home. Newborn Screening: A Blueprint for the Future* [National Newborn Screening and Genetics Resource Center, 2000]; and (3) *Integrating Genetic Services into Public Health—Guidance for State and Territorial Programs from the National Newborn Screening and Genetics Resource Center* [Kaye et al., 2001].

Healthcare Provided by Genetics Specialists. The narrowest level of the framework includes services provided by individuals trained in genetics such as medical geneticists, genetic counselors, and nurses and dietitians with specialty genetics training. Services provided by these individuals might include diagnosis, counseling, care coordination, or treatment and management. This area also includes genetics laboratory testing for individuals including molecular genetics, biochemical genetics, and cytogenetics.

Genetics Services Outcomes Menu

Work group members were aware of other activities to improve evaluation of newborn screening laboratory and short-term follow-up services, the InheritQual project and a joint project of the American College of Medical Genetics and the Mountain States Genetics Regional Collaborative Center to develop process quality indicators for evaluating clinical genetics services and therefore decided not to emphasize these areas in further developing outcomes for the menu.

Several options were considered for organizing the Menu: the *Defining Genetics Services Framework*; the main concept themes which emerged from content of the outcome areas; and a logic model framework of short, medium, and long range outcomes, phrased in terms of changes in knowledge or understanding, behavior, or society. As with the *Defining Genetics Services Framework*, a hybrid organization scheme was chosen as the most useful: knowledge and information; financing; screening and identification; diagnosis, treatment, and management; and population health (Fig. 2).

Public health programs are often evaluated for their effectiveness in accomplishing one or more of the three levels of disease prevention: primary, secondary, or tertiary. Definitions of primary, secondary, and tertiary prevention vary in the literature. Generally, primary prevention is defined as preventing the occurrence of a disease. On a population level this is usually measured in terms of change in incidence of a condition. The work group decided not to include outcomes related to change in incidence of genetic conditions in the Outcomes Menu for two reasons. First, group members agreed that framing outcomes in terms of decreasing incidence might be seen as devaluing the lives of people living with genetic conditions. Second, many genetics specialists believe their role is to assist people in making decisions consistent with that person's values and beliefs rather than promoting a particular course of action such as avoiding conception for couples at risk for having a baby with a genetic condition or terminating a pregnancy if a fetus has a genetic condition.

Generally, secondary prevention means preventing health problems due to an existing condition. Tertiary prevention means preventing disability that might result from an existing condition and maximizing quality of life. Representatives from all constituencies were interested in including outcomes aimed at secondary and tertiary prevention. Outcome V.A.2, "Symptoms or complications of genetic conditions are prevented or detected early" is an example of secondary prevention. Outcome V.B.3, "Individual and family daily functioning is optimal" is an example of tertiary prevention.

DISCUSSION

Inclusive Group Process

From the outset, the work group viewed the diversity of its members in terms of constituency represented and experience with the topic as a strength and believed that including clinicians, family

advocates, and public health professionals in the development process would lend credibility to its products. Values of the various constituencies are reflected in the Outcomes Menu. For example, family advocates felt strongly that outcomes should be written in a positive manner whenever possible and almost all outcomes in the Menu are framed in this way.

Using the Defining Genetics Services Framework

The *Defining Genetics Services Framework* articulates the relationship between population-targeted public health and individual-targeted personal healthcare activities. Work group members reported that it enhanced their understanding of the perspectives of collaborative participants from other constituencies. We foresee the framework being used in multiple ways. For example, state genetics programs might use the framework to orient new advisory committee members or allocate staff time or funding. Family or consumer organizations might use the framework to help target educational initiatives. Researchers might use the framework to study aspects of key relationships between public health infrastructure and personal health genetics services.

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Using the Genetics Services Outcomes Menu

We envision the *Genetics Services Outcomes Menu* being used as the term "menu" implies—users choose outcomes from the menu depending on their own purposes. Menu outcomes can also be modified. Several contain multiple permutations of a single concept, as in outcome I.B.1.—"healthcare providers use current information about incidence; prevalence; epidemiology; diagnosis; and treatment of genetic conditions to prevent, cure, and treat individuals with heritable conditions." The work group considered whether to simplify these outcomes, but decided it would be more useful to convey all possible permutations. Users can simplify the outcome if desired. For example, outcome I.B.1. could be simplified to "healthcare providers use up-to-date information to care for their patients with heritable conditions." Users might also want to rephrase the outcomes as changes rather than achieved results. For example, outcome I.B.1. could be modified to "increase in the proportion of healthcare providers who use up-to-date information to care for their patients with heritable conditions."

The *Defining Genetics Services Framework* and the *Genetics Services Outcome Menu* were used to update the WSGSC Regional Plan. Activities in the plan and their corresponding outcomes were reorganized to reflect the Outcomes Menu categories of knowledge and information; financing; screening and identification; diagnosis, treatment, and management; and population health; and modified versions of several of the outcomes were added to the plan.

We hope state genetics programs and others will try out these tools to determine their usefulness to their own programs, and share their experiences.

Next Steps

The *Genetics Services Outcomes* Menu is intended to be a fluid document. The WSGSC plans further work on developing health outcomes and indicators for the genetics healthcare services delivery system and direct healthcare levels of the *Defining Genetics Services Framework*.

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