

**COLORADO DEPARTMENT OF PUBLIC HEALTH
AND ENVIRONMENT**

STATE GENETICS SERVICES PLAN

February 18, 2002

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- C) *Colorado Vital Statistics Summary by County and Region, Colorado Residents, Annual Average, 1996-2000*. Demography section, Colorado Division of Local Government. <http://www.cdphe.state.co.us/hs/summaryweb2000.pdf>
- D) *Public and Community Health Nurses Survey, 2000*. Colorado Department of Public Health & Environment.
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I. ACKNOWLEDGEMENTS

A) The State Genetics Advisory Committee

The development of a rational, inclusive approach to integrating genetics into all appropriate areas of public health is a monumental undertaking. Making every effort possible to address all areas of concern, need and opportunity requires the input and perspectives of representatives from all areas of public and private healthcare, including those representing the end user and consumer perspectives. The Colorado Department of Public Health and Environment (CDPHE) therefore has assembled a State Genetics Advisory Committee (GAC), comprised of a diverse group of medical professionals, public health officials, scientists, academics, policy and legal advisors, and consumers. Their input, oversight, review and suggestions continue to be of critical importance and value to the development of a State Plan for the Provision of Genetic Services which will effectively coordinate and strengthen Colorado's activities in genetics and public health.

Members of the GAC serve two-year, renewable terms. All service is voluntary; administrative support of their efforts is provided through the State Planning Grant.

B) Federal and Statewide Agencies and Boards

True to the need for collaboration, coordination and cooperation between various federal and state agencies in the development of public health programs, multiple human and health services agencies are continuously interfacing with CDPHE in order to create synergy, support and economies of scale as new programs are brought to the public. The existing and evolving genetics programs that form the foundation for this report draw upon invaluable communication and shared information between the Plan developers and the Disease Control and Environmental Epidemiology Division, which tracks, controls and prevents communicable diseases and other conditions in Colorado to reduce illness and premature deaths. This division also assesses risks from toxic exposures in the environment to prevent adverse health effects, a function projected to be seminal as genetic testing expands and becomes available for segments of the population beyond the current focus on teratogens and potential birth defects.

Pertinent to current genetic programs and development of enhanced services to reduce birth defects and improve child health, the State Genetics Services Plan also is indebted to the use of population data and continuing services reviews from multiple sources, including the Colorado Department of Public Health & Environment's Healthy People 2000 report, and many others, which are discussed in detail in **Section F**. These programs include the Colorado Responds to Children with Special Needs (CRCSN); the Centers for Disease Control and Prevention (CDC) and four other states to determine the prevalence of fetal alcohol syndrome (FAS), from which information is correlated with data regarding incidence of developmental disabilities and long term patient management issues in order to plan for services and evaluate the effectiveness of prevention efforts. CDC programs to collect, analyze and make available data on birth defects, operate regional centers for applied epidemiologic research on the prevention of birth defects, and inform and educate the public about the prevention of birth defects also are correlated into this needs assessment report.

Many of the Prevention and Intervention Services for Children and Youth Division (PSD) programs are funded under the federal Title V grant (see **Section F (3-a)**). Of particular value to the development of this plan is the ongoing coordination of activities, data, resource monitoring and education which are the cornerstone of CDPHE activities through the Mountain States Genetics Network (MSGN). This organization is made up of genetic service providers and consumers from six Rocky Mountain States, Arizona, Colorado, Montana, New Mexico, Utah, and Wyoming.

Finally, this assessment and plan benefits greatly from review of current adult services through CDPHE Health Promotion and Disease Prevention Division Cardiovascular Health, administered through the Chronic Disease Section of the Colorado Department of Public Health and Environment.

II. EXECUTIVE SUMMARY

Understanding genetics and its impact on all segments of the population helps to clarify the enormity, and the necessity, of developing an extended plan for the expanded integration of medical genetics into public health programs.

Some genetic information can contribute to good health. Therefore, understanding genetics and the value of genetic information allows us to diagnose and sometimes treat deleterious mutations (i.e. develop patient profiles and optimize patient management programs). In other words, identifying deleterious mutations that cause a predisposition to disease can sometimes provide options for avoidance or intervention. Just as importantly, identifying deleterious mutations that predispose a carrier to passing on a deleterious mutation to offspring allows individuals to make informed reproductive choices.

Emerging information about genetics indicates that this area of medical science will have far-reaching impact on health care, and consequently, on public health programs. A major consequence of the Human Genome Project and other genetics research has been the mounting evidence some genes (“deleterious mutations”) make people unhealthy, and that birth defects and chronic diseases such as cancer, cardiovascular disease, dementia and mental health disorders, diabetes, obesity, blood and immune disorders all have major, identifiable genetic risk factors that can act in concert with personal behaviors and other environmental risk factors to cause disease in virtually all segments of the population.

It is the mission of the State Genetics Services Plan to integrate genetics into health care in order to develop public health interventions that will realize fully the goals of Healthy People 2010 through the seamless integration of quality genetics services into health care systems in Colorado.

To improve outcomes, and when possible prevent morbidity and mortality among Coloradoans with or at risk of genetic, congenital and/or hereditary disorders by assuring the continued review, assessment, integration and delivery of genetic services to all individuals and families who may be at risk. This will include addressing various underserved segments of the population, including low income and ethnic groups, women and seniors currently unaware of, or without access to, evolving genetics-based health care services.

Previously focused on specific disorders affecting relatively small percentages of the population, advances in genomics continue to significantly broaden the definition of “at risk” populations. Given the inclusive nature of genetics as the cornerstone of an individual’s medical profile, the state plan proposes to accomplish the following:

1. Utilizing an age-based approach, define goals according to target demographics: preconception, newborn, young children, adolescents, adults and seniors.
2. Review genetic services currently available to these demographics.
3. Determine current gaps in genetics services to these demographics.
4. Establish priorities for policy development/service delivery by age group:
 - a. Current gaps
 - b. Short-term needs
 - c. Long-term needs
5. Make recommendations for addressing priorities.
6. Determine the State’s role for addressing priorities.
7. Establish collaborative opportunities.
8. Establish barriers to delivery.
9. Create timeline.

In order to deliver the Plan recommendations in a more accessible format, a matrix has been designed, which when fully developed, will hopefully provide state policy makers and influencers with the information necessary to make informed decisions to act. Meeting long term goals will eventually involve developing protocols for the creation of inclusive programs to routinely update existing public and private health programs. As with all aspects of the Colorado State Genetics Services Plan, the matrix will be a constantly evolving document which will reflect advances in genetics medicine and explore new options to improve the health of the population.

III. DEFINITION OF PURPOSE

A) Background

The challenges facing public health at the dawn of the 21st century reflect a decidedly daunting combination of changing opportunities, ethical ambiguity and economic realities. While the fundamental mission of public health - i.e. “fulfilling society’s interest in assuring conditions in which people can be healthy” – hasn’t changed, the definition and means of accomplishing the mission have become complicated by rising costs in services provision, managed care, dramatic advances in medicine, and complex ethical issues which in many cases must be addressed on an individual basis.

Much of this controversy is focused within the field of medical genetics, where costs, access, service provision and ethics collide as science and technology rapidly outpace existing standards of care.

Further complicating the situation is the general awareness among the professional and public sectors regarding the daily impact genetics is making on medicine and health. Rapid advances in genetics are continually providing new insight for disease prevention and health promotion, with applications for chronic and infectious diseases.¹ As a result of the Human Genome Project, exciting discoveries regarding cancer, cardiovascular disease, birth defects, mental health disorders, diabetes, immuno-deficiency disorders and other conditions are creating expectations and driving demand for services.

The health benefits of the use of genetic testing and eventually, genetic therapies, are undisputed. Genetic tests can save costs by identifying those in high-risk families who might benefit from close medical surveillance. Conversely, costs of surveillance, not to mention the costs of personal stress, can be saved by identifying those not at increased risk.

There are currently approximately 450 genetic tests available, most for disorders which are relatively rare. A large number of these tests are used in maternal health and newborn screening programs. But new genetic markers for relatively high incidence disorders for both children and adults, such as hereditary hemochromatosis (HH), a common, treatable, disorder that affects approximately 1 in 400 individuals of Northern European descent, are rapidly being developed. And the number of studies investigating the genetic basis of other diseases is growing rapidly, as are new applications of genetic information. For example, pharmacogenomics, a term coined in recent years, is another genetic medicine advance that allows for better treatment of disease based on an individual's genetic make-up and possible gene-drug interactions.

Although it is generally projected that genetics will soon play a role in nearly every aspect of healthcare, current provision of services is relatively limited, and the timeframe for the broad application and adoption of genetic medicine is unpredictable on all fronts. The questions that are emerging from the availability of genetic tests will make new demands on public health systems, and will directly impact the specific development of the Colorado State Genetics Plan. These questions include:

- What criteria should public health use to determine whether a genetic test should be recommended?
- How should genetic tests and services that are not population-based be incorporated into standards of care?
- Should public health only recommend screening programs for treatable disorders?
- Is the primary role of the CDPHE provide information and education only on services currently available through public health programs? Should it also provide educational materials on emerging medicine and enhanced standards of care? What responsibility must CDPHE then take for potentially increasing demand for services that are too expensive and too specialized for the general population (i.e. only available through highly specialized labs)?
- As genetics drives new standards of care, how will new tests and therapies be funded?

¹ Association of State and Territorial Health Officers (ASTHO). PHG-1 Public Health Genetics Policy Statement 2001.

- Can legislation truly guarantee non-discrimination among insurers and employers as more prognostic genetic health information becomes available?
- Will increased genetic testing widen the gaps between general and underserved populations?
- How will the State address the issues surrounding chronic adult care and long-term patient management for chronic disorders predicted by genetic testing?

B) What is the Responsibility of Public Health?

a. Assessment

To improve health it is important to assess the relationships between genetics and health including how genes interact with our surrounding environments. According to the book, *Genetics and Public Health in the 21st Century*, surveillance is needed to determine:

1. the population frequency of genetic variants that predispose people to specific diseases, both common and rare;
2. the population frequency of morbidity and mortality associated with such diseases; and
3. the prevalence and effects of environmental factors known to interact with given genotypes in producing disease.²

Genetic screening eventually could replace traditional screening methods for certain diseases and may become the only screening method for other diseases. Vital to implementing any guidelines is the assessment of a variety of factors common to public health. These factors include: the availability of quality genetics resources in the community, the appropriateness of genetics technologies offered to the community, the accessibility of clinical and laboratory services, the cost benefit of using genetics technology, and the community's knowledge of the use of genetics to improve health. The assessment of these factors alerts health officials and others to areas in which policies should be developed and for which better assurance of services is needed.

Developing short and long term programs to improve the public health and better facilitate the collaborative partnerships between local and regional public agencies and the private sector will require ongoing assessment of current resources and programs. The assessment process as it applies to development of a State genetics plan also requires review of current population data, service availability and support resources, and benchmarking against established performance measures.

It is also incumbent upon the CDPHE to create a cycle of reassessment and planning to identify new priorities, emerging need, issues and obstacles to healthcare services delivery, and gaps in services which could undermine the goals of the Plan. As noted, genetics services are evolving daily, and the rapid aging of any needs assessment will be mirrored in lagging services and underserved publics.

² Koury et al, 2000.

b. Policy Development

A primary goal of the State Genetics Advisory Committee and the CDPHE in the development of a State Genetics Services Plan is to develop a practical approach to creating policy that will serve the Colorado population in the delivery of genetic services. The development of public health policies ensures that the public can access safe, effective and quality services without unnecessary apprehension. Public health policies can also provide the public with objective guidance and information to empower the decision-making regarding the use of genetic technologies.

The development of good genetics policies requires input from a broad-based spectrum of disciplines, professional backgrounds, interest groups, stakeholders, and consumers. There are several issues for which policy is need to ensure the public's health and minimize potential harm including health insurance discrimination, population screening, and privacy and confidentiality.

But while obvious in need, the form and process to actually finalize new policy for integrating emerging genetic technologies into public health is both laborious and daunting. Existing programs, limited funding options, special interests and complex bureaucracies all must be projected into the development of a model that will provide the best course of action to achieve positive results in the best interests of the public health. While all mechanisms have not yet been identified to achieve this goal, the guidelines for moving forward include the following policy advisements, which will be incorporated into this plan and into the activities of the Genetics Advisory Committee.

- Provide advisement to the state and other CDPHE programs regarding the evolving genetics program, philosophy and policies, and how these affect individuals and families, emphasizing primary, secondary, and tertiary prevention, management, and treatment of disabilities caused by genetic factors.
- Review, comment on, and recommend legislation pertaining to human genetic issues.
- Promote (advocate) the development and implementation of genetic health services programs, education, and policy.

c. Assurance

Public health agencies assure their constituents that services necessary to achieve goals are provided, either by encouraging action by other entities (private or public sector), by requiring such action through regulation, or by providing services directly (IOM, 1988). Agencies may collaborate with other public and private entities and educate public health and private health care workers about the use of genetic information to improve health. Assurance that the best interests of the public health in regards to the delivery of genetics services are met will require dedicated collaboration between CDPHE and its many public and private partners. Agencies may collaborate with other public and private

entities and educate public health and private health care workers about the use of genetic information to improve health. To assure that access to services and quality control are maintained for all uninsured/underinsured Coloradans, CDPHE must identify strategies to assure there are a sufficient number of public health workers to address the needs of our citizens, or determine practical means to share services regionally and through private providers. Finally, CDPHE must work diligently to assure that public dollars are invested properly by continuously evaluating the cost-effectiveness of personal and population-based health care services. To accomplish this, part of the assurance process must rely on the reassessment function of the State Genetics Services Plan, including

- Regular consultation with Genetic Services Section of the CDPHE on contemporary issues regarding patients and/or families with genetic health care needs.
- Consultation with the Genetic Services Section on contemporary issues regarding providers of health and social services, with particular emphasis on providers of genetic health care services.
- Promotion of family-centered, community-based, culturally competent, comprehensive, coordinated social and health care systems to meet the genetic health care needs of clients and families.

Programmatically, the incorporation of up-to-date genetic information in areas such as maternal and child health, occupational health, and prevention and disease focused programs will improve outcomes by providing better prevention information. Data systems capable of monitoring the quality of individual services will need to include genetic information, as this data may present opportunities for targeted prevention. Outcome evaluations that include genetic information will create an opportunity to develop more effective policies and practices. Additionally, health agencies will need to assure the availability and quality of laboratory and clinical services.

C) Areas of Focus

Although each new area of genetic testing and disease management will generate its own issues, it is clear that basic guidelines for changes in standards of care, models for policy development, methods of service delivery and provision for follow-up services are vital to the timely delivery of new public health initiatives. To this end, CDPHE must take care that its initial areas of focus are appropriate to the development of foundational models, which will provide the infrastructure for continuing evaluation and change as the field of genetics expands and new services become available.

For the purposes of this document, in order to create the basis for a state genetics plan for Colorado, several specific areas of focus therefore have been identified and will be discussed in the context of plan and policy development. They include:

- Gaps in current healthcare services in light of rapidly emerging genetic technology.
- Gaps in follow-up strategies for long-term care, prevention and intervention monitoring and data collection.
- Limited infrastructure building models and services to accommodate genetic healthcare issues that potentially affect the entire population. These include

development and maintenance of health systems, standards and guidelines, training, data, and planning as they apply to assessment, policy development and assurance efforts.

- Gaps in current provision for adults with chronic disorders.
- Gaps in current and future provision for special populations and cultural competencies.
- Barriers to services and care due to limited professional resources and staffing.
- Barriers to services and care due to limited funding resources.
- Issues regarding third-party reimbursement with State contractors for specialty services.
- Overwhelming public education imperatives in light of the complexity and pace of genetic discovery.

IV. NEEDS ASSESSMENT

K) Introduction

The growing impact of genetics on healthcare has given rise to a number of issues which are increasingly the concern of the State. The potential for early diagnosis, predictive testing and new therapies as a result of advances in medical genetics has created new challenges to address how these services should be incorporated into evolving standards of care, and what role public health systems should play in the process. As steward of the public health, the Colorado Department of Public Health and Environment is obliged to determine the current and projected needs of the public from both services delivery and education perspectives. Additionally, in order to accomplish this, the State must also be actively involved in understanding and supporting ongoing professional education in the field of genetics in order to oversee policy development and assure quality service delivery in the context of public health.

Furthermore, the issue has become increasingly complex in light of the progress of sequencing the human genome. The Human Genome Project has potentially opened the door to a new era in healthcare, in which predictive diagnostics and gene therapy offer new hope that both inherited disorders and common diseases including cancer and heart disease will be conquered through genetic medicine.

Consequently, Colorado is proceeding to formally begin development of a long-term program to address the issues surrounding the delivery of genetics services to its population. Following standard guidelines, the State will assess needs, create recommendations for the development of appropriate policy and provide assurance review to monitor service delivery.

1) Review of Other State Programs To Date

Colorado is not the first state to see the need to address these issues. In development of this document, several plans from other state departments of health were reviewed, including state genetics plans funded by federal planning grants for Arizona, Hawaii, Ohio and Washington. In brief, these plans included the following key points:

Arizona

Exhaustively researched over three years and completed in 2000, with inclusions of general information conducive to population education rather than specific plan development. Much of the document contains a literal review of existing programs and the history and process of the development of the document, rather than containing this information in addenda to the actual plan as resource, contributory or substantiating data. Key components include

- Well-organized Project Approach
- Poll of major concerns of genetics professionals
- Current challenges to delivery of existing genetics services

Utility

- Final Plan Recommendations and Action Items only provided for the development of program components, but no plan to develop the components.
- The final plan was essentially a discussion of what the State needs to do, not how to do it.
- Plan had few forward-looking recommendations. Nearly all Action Items were geared towards currently provided programs and needs without provision for expanded services based on evolving technology.
- There were no expanded reimbursement or delivery system strategies.

Hawaii

The Hawaii Plan presents a direct, well-organized approach that indicates a sound knowledge of the current services and population needs. The Long-term Plan, developed in 1996, addresses only prenatal genetics services, and assumes no additional funding for expanded programs. Needs assessment data was carried forward from 1993.

Key Components

- Presentation of current service levels (at the time) indicated an unusually well developed integration between public and private health systems.
- Legislative support of prenatal genetic screening services, including funding, is already in place.

Utility

- The final plan was essentially only a discussion of what the State needs to do, not how to do it.
- Plan had very little forward-looking data. Nearly all Action Items were geared towards currently provided programs and needs without provision for expanded services based on evolving technology, and adult and chronic disease issues.
- No expanded reimbursement or delivery system strategies.

Ohio

Ohio's plan was written in narrative style, organized more as a public information brochure than a State Genetics Plan. The document contained no significant original program components. There was no development of primary research or needs assessment data.

Key Components

- Approachable case studies may have value to consumers at an introductory level.

Utility

- None from a practical program implementation perspective

Washington

Highly intelligent and well researched, the Washington State Genetics Plan, completed in 1997, provided an excellent framework for implementation of a workable program. The methodology, organization and analysis of the needs assessment data were impressive. The Plan's only weakness is in its failure to address genetics services beyond maternal and child health populations.

Key Components

- Good survey development.
- In most cases, survey samples of professional and consumer segments were large enough to be statistically relevant.
- Thorough review of all population segments

Utility

- 3-5 year Plan addresses all key issues, including reimbursement, quality assurance, consumer and professional education.
- Recommendations are practical and realistic.
- Organization of Plan in matrix form makes the Action Items clear and accessible.

2) Projected Demand and Genetic Services Delivery Assessment

The current status of medical genetics research and patient applications can be projected to have far-reaching effects upon consumers and the CDPHE. According to Francis Collins, M.D., Ph.D., director of the National Human Genome Research Institute at Jackson Laboratory, Bar Harbor, Maine, the next decade will lead to the discovery of the common gene variants for common diseases; predictive tests will be available for at least 25 disorders, with intervention available for most of these conditions; gene therapy will continue to progress and prove successful for several conditions; preimplantation diagnoses will be more widely available; and primary care providers will practice genetic medicine.

Previously viewed as necessary services for special needs populations only, the expansion of various types of genetics services will be required to support a larger portion of the population. Access to clinical services, including genetic counseling, genetic testing,

prevention and patient management strategies will need to be provided not only for maternal and child health programs, but also for adults with various genetic concerns, including cancer, hemochromatosis, clotting disorders, genetic neurological diseases, chromosomal disorders, connective tissue disorders, and other diseases in a list of health issues with a strong genetic component that is growing exponentially. Consequently, this needs assessment includes:

- Assessment of the approximate percentage of the population that might be eligible and/or require genetic services. This percentage would include both direct and indirect recipients of clinical services, including patients and their extended families.
- Assessment of the maximum delivery capacities for existing services providers, both public and private.
- Assessment of the need to determine alternative or additional options for delivery of services through new delivery channels.

Justification for the project is based on the following assumptions:

- Advances in medical genetics will continue.
- Coverage by the media of subjects relating to genetics and healthcare will continue, providing consumers with more information which is projected to generate increased interest in, and demand for, genetic education and services.
- Primary care physicians and public health nurses will be the “first line” contacts for patients and the general public to answer questions and identify need for services.

B) PURPOSE

The following Needs Assessment document has been prepared to support the development of a 2001 Colorado State Genetics Plan that will adequately serve the immediate and long term needs of the population. The assessment, as the initial development component of the State Genetics Plan, takes into account Colorado state geography and population demographics; existing genetics-related public health resources; projected need for services by 2005; and current data collection systems. This data was then reviewed in the context of emerging medical genetic technologies that will directly impact healthcare among general and special needs populations.

This assessment provides a “snapshot” of the Colorado health services environment as it pertains to genetics. Ultimately, the goal of this assessment is to validate and guide the development of a state genetics plan to adequately service the Colorado population in the next five years. By reviewing current population statistics, service availability and developing technologies which will directly impact the provision of additional services in the near future, Colorado will be better equipped to develop a relevant State Genetics Plan. This plan will address strategies to overcome current and projected gaps in, and barriers to, quality genetics services for all segments of the population.

C) METHODOLOGY

In order to fully understand the projected need for genetics services in the next five years, it is critical that current statistics and service availability be reviewed in light of the rapid advancement of genetics technologies and their direct impact on the practice of medicine. This needs assessment reviewed and correlated foundational data currently available through multiple resources including the Health Statistics and Vital Records Section (HSVR) of the Colorado Department of Health, the National Human Genome Research Institute of the National Institutes of Health, the Centers for Disease Control and Prevention, the National Center for Biotechnology Information and the Mountain States Genetics Network. Each of these resources provides access to seminal information for public and professional use.

Additionally, the following secondary materials have been reviewed and analyzed to provide supplemental perspective in the development of genetics programs as a result of this assessment:

- State Genetics Programs for Arizona, Hawaii, Ohio and Washington (see above).
- Current Genetics Educational Materials from the Colorado Department of Health and Human Services.
- Recent discussions of genetics in professional and consumer media (newspapers, magazines – see *Bibliography* below).
- Current genetic testing availability and projections for new testing.
- Numbers of primary care physicians and public health nurses currently servicing the Colorado population.
- Existing Genetics-related Public Health Systems
 - i. Existing Genetics Services Resources
 - ii. Projected Need for Services by 2005
 - iii. Reviews
 1. List of current resources (Public and Private) per the Mountain States Genetics Network and the national *GeneClinics* listing from the University of Washington, Seattle.
 2. Review of Public and Community Health Nurses Survey Results

D) STATE SIZE AND GEOGRAPHY

Geographic Barriers to Service Delivery

Bisected longitudinally by the Rocky Mountains, Colorado is the seventh largest state in the continental U.S., covering 103,600 miles. The geography presents multiple factors which influence the efficiency and equality of health services delivery. The majority of the population, which is expected to reach more than 4.6 million by 2005, is concentrated in nine counties, which hold more than 75 percent of the Colorado populace and support the majority of the State's economic activity in, or near, urban areas.

Growing in population by more than 1 million between the 1990 and 2000 Census, the most recent birth rate shows an unprecedented 18.8 percent increase, adding more than 12,000 individuals to the population in 1999. A strong employment pool and low unemployment of approximately 2.6 percent (January, 2000), coupled with a highly admired quality of life, has drawn significant migration to both urban and rural areas.

**Colorado State Topography,
with bisecting mountain range detail:**



However, many of the new jobs are in the vibrant tourism industry, which primarily creates jobs in relatively remote mountain resort areas—the Front Range area--the majority of which are two-to-four hours from major medical facilities offering genetic services.

Of the State's counties, 31 are classified as "frontier counties, containing less than six people per square mile. Many of these sparsely populated counties are part of the mountainous "Western Slope" (referring to the western slope of the continental divide), which has more than 54 peaks over 14,000 feet high, creating significant physical barriers to major urban areas with full medical facilities.

On the eastern side of the Rocky Mountains, broad, flat prairies support dozens of small farming communities with populations challenged by long distances separating them from major medical centers. There are particular limitations to the delivery of genetic services in these rural areas, as the great majority state-of-the-art services are located in the Denver-Colorado Springs urban clusters, which sit on the east side of the Rockies. Regional clinics support most basic public health needs. However, the paucity of qualified specialists to address genetics-related maternal and child health needs, not to mention adult services for late onset disorders with a genetic component, creates a significant gap in the state's ability to service the population.

These disparities have particular significance among low income segments of the population, which also often reflect racial and ethnic groupings. Particularly in the identification and treatment of genetic disorders among these groups, the state is challenged to meet even current standards of care, making the addition of new genetic services extremely problematic.



E) POPULATION DEMOGRAPHICS

1. 2000 State Census

The U.S. Bureau of the Census released its April 1, 2000 count of the State's population on December 27, 2000. The number, 4,301,261 people, makes Colorado the 24th most populated state in the country. The 2000 census number of 4.3 million amounts to an increase of more than 1 million people (30.6%) since the 1990 population count of 3.2 million. This equals an annual average growth rate of 2.7 percent, which compares to an average of 2.3 percent implied by the Census Bureau's estimates during the decade.

The migration of new population to take advantage of the stable job market has resulted in a decrease in the state poverty level, which decreased from 11.7 percent in 1990 to 10.5 percent in 1996. This translates to 14.6 percent of children under 18, or roughly 180,000 children living below the federal poverty line. The numbers also indicate that approximately 100,100 women of childbearing age also fall into this category. A majority of these numbers are located in the southern counties..

Despite the general state growth and prosperity, then, it is clear that a significant portion of the current population is challenged to receive full access to many medical services, including genetic services. Additionally, there are profound disparities in the health status of various segments of the population, particularly among racial/ethnic segments. This is particularly obvious in review of maternal and child health issues associated with the growth of the Hispanic community, the birth rate of which increased by more than 57 percent from 1990 to 1998. These are discussed below as part of the vital statistics review.

2. COLORADO STATE VITAL STATISTICS

The attached *Colorado Vital Statistics Summary by County and Region, Annual Average (1996-2000)* provides summary information relevant to projected genetics services issues, primarily due to the expanded scope of applications for various genetic tests expected to become available in the near future.

In brief, the scope of involvement that advances in genetic technology are likely to have in public health issues can be quantified by a review of the key statistics:

Live Births – 59,881 live births reported; birth rate of 14.6 per 1,000 population; general fertility rate of 63.6 per 1,000 women age 15-44. These statistics have already been impacted by improvements in prenatal screening and in vitro fertilization techniques.

Infant Deaths – 396, infant mortality rate 6.6 per 1,000 live births. Advances are expected to continue in early screening techniques which will also allow for intervention in difficult pregnancies, thereby decreasing infant mortality.

Crude Death Rates for Ten Leading Causes of Death in Colorado – Six of the ten leading causes of death in the state – heart disease, cancer, cerebrovascular disease, diabetes, atherosclerosis, Alzheimer’s disease – all have genetic components which have current and projected genetics services applications, including early detection and intervention.

Review of the data sets reveals many disparities related to special populations. Some examples of the statewide county data are included here, or follow in Appendix C, or can be viewed in their entirety at

<http://www.cdphe.state.co.us/fc/mchdatasets/mchdatahorn.asp>.

The greatest amount of data focuses on maternal and child health issues, which bear a direct relationship to the perceived need for access to genetic services that address these issues. A full review of the data is available on the Colorado Department of Public Health & Environment’s Healthy People 2000 website, which provides specific descriptions of the Healthy People 2000 objectives and areas of concern, including low birth weight, lack of immunization among children, and obvious health disparities among racial/ethnic groups. These differences suggest differential access to health care, sometimes revealing patterns related to socioeconomic differences.

Beyond the maternal and child health programs (further outlined below), there are relatively few services geared towards adults, whether those dealing with disorders from birth, or those with later onset disorders. Further, there are no consistent protocols or provisions for predictive genetic counseling and genetic testing for genetics related diseases such as cancer. These areas in particular are expanding rapidly, and will demand attention from the State in the very near future.

F. Existing Genetics-related Public Health Resources

As state agencies charged with the development of public health programs, multiple human and health services agencies are continuously interfacing with CDPHE in order to create synergy, support and economies of scale as new programs are brought to the public. The existing and evolving genetics programs that form the foundation for this report draw upon communication and shared information between the Plan developers and the Disease Control

and Environmental Epidemiology Division, which tracks, controls and prevents communicable diseases and other conditions in Colorado to reduce illness and premature deaths. This division also assesses risks of exposure to toxins in the environment to prevent adverse health effects, a function projected to be seminal as genetic testing becomes available for segments of the population who may be at greater risk from exposure to teratogens due to certain genetic risk factors.

Pertinent to current genetic programs and development of enhanced services to reduce birth defects and improve child health, the State Genetics Services Plan is using population data and continuing services review from the Colorado Responds to Children with Special Needs (CRCNS), Colorado's birth defects monitoring and prevention program. The purposes of CRCNS are to maintain a database of young children with birth defects, developmental disabilities, and risks for developmental delay; to provide statistics to other programs, agencies and researchers; and to prevent birth defects and related disabilities by linking children and families with early intervention services.

Data used to assess population needs is also gathered from the Centers for Disease Control and Prevention (CDC) and four other states to determine the prevalence of fetal alcohol syndrome (FAS). This information is correlated with data regarding incidence of developmental disabilities and long term patient management issues in order to plan for services and evaluate the effectiveness of prevention efforts. Fetal alcohol syndrome is a common cause of mental retardation and may account for as much as eleven percent of residential care for mental retardation in the United States. Other alcohol-related birth and neuro-developmental defects can result in lifelong physical, behavioral and cognitive abnormalities.

CDC programs to collect, analyze, and make available data on birth defects, operate regional centers for applied epidemiological research on the prevention of birth defects, and inform and educate the public about the prevention of birth defects also are correlated into this needs assessment report.

1) Colorado State Genetics Programs

Of particular value to the development of this plan is the ongoing coordination of activities, data, resource monitoring and education which are the cornerstone of CDPHE activities through the Mountain States Genetics Network (MSGN). This organization is made up of genetic service providers and consumers from six Rocky Mountain States, Arizona, Colorado, Montana, New Mexico, Utah, and Wyoming. Over two hundred members include physicians, geneticists, cytogeneticists, molecular biologists, genetic counselors, genetic nurses, public health officials and persons affected by genetic conditions and their families. The Network's mission mirrors that of the State Genetics Plan in regards to the following"

- to assess the need for genetics services throughout the region
- to establish and maintain a database of genetic services provided in the region
- to promote collaboration and the sharing of resources among genetics professionals throughout the region

- to promote cultural sensitivity and consumer participation in genetics service issues
- to develop and carry out genetics education for primary care and other health care providers
- to assist member states with integrating genetics services into their maternal and child health programs
- to monitor the quality of clinical and laboratory genetics services within the region
- to collaborate with the Council of Regional Networks in addressing public health genetics issues at the state and national levels
- to measure the impact of managed care on genetics services within the region and to act to assure comprehensive genetics service access to all

2) Direct Genetics Services

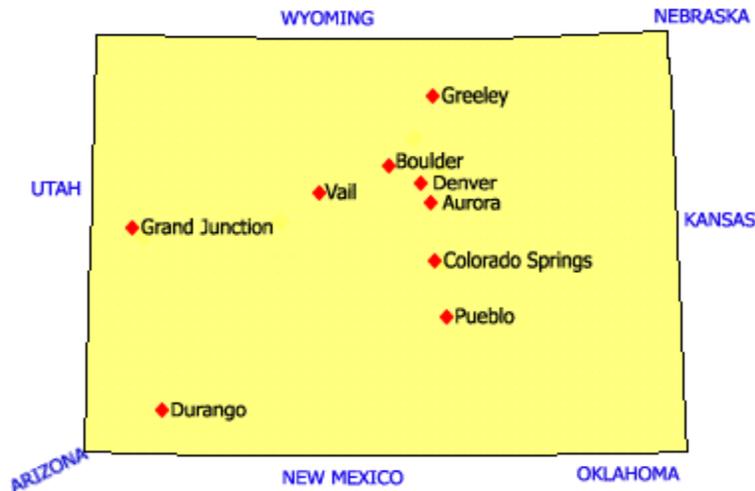
The following map indicates locations for public and private genetics services. These services included clinical services, laboratories and counseling facilities. They include:

- Women’s Health Services, which includes the Prenatal and Prenatal Plus Programs;
- MCH Medical Consultant & Genetic Services, which includes Newborn Screening;
- Health Care Program for Children with Special Needs, which includes the Development & Evaluation Clinical Program, and the Newborn Hearing Screening Program.

A full listing of all locations is provided in the index. Predictably, the majority of these services are concentrated in urban areas, primarily in the greater Denver and Colorado Springs areas. This situation causes reduced opportunity to provide special services such as genetics to the rural population. With more than 1.1 million individuals currently living outside of the high population centers, genetics services are likely to be provided primarily by referrals from primary care physicians and public health extension services.

Colorado State Genetics Service Locations

COLORADO



Genetic counseling services are provided throughout the state of Colorado, primarily by the Genetics Unit of the University of Colorado Health Sciences Center and its affiliate, The Children's Hospital. These activities are performed under contract with CDPHE which carries out consumer support activities, newborn screening laboratory services and subsequent follow-up. Services currently are available for adults and children and include general genetic evaluation and counseling, prenatal diagnosis, and single gene counseling and management. Outreach clinics are currently held in Colorado Springs, Durango, Grand Junction, Greeley, and Pueblo. Physicians, nurses, schools, and other health professionals can make referrals by calling the appropriate phone number. Self-referrals are also accepted. Laboratory services include cytogenetics, molecular genetics, and biochemical genetics. A variety of private genetic services are also available, including prenatal diagnosis and in vitro fertilization.

a) Listing of Genetic Service Providers in Colorado

(See Addendum A for a complete listing of genetics services in Colorado.)

3) Women's and Children's Services

Current state genetics-related programs are primarily serviced under the Maternal, Infant and Child Health program. These programs, which include diagnostic screening, counseling and education components, provide for prenatal screening for open neural tube defects and Down syndrome; newborn screening for metabolic and certain genetic disorders including sickle

cell anemia, phenylketonuria, galactosemia, biotinidase deficiency, cystic fibrosis, congenital hypothyroidism and congenital adrenal hyperplasia; statewide newborn hearing screening and child health screening and program support for developmental and perceptual disorders, including deafness. *The State's Newborn Metabolic Screening Program* tests approximately 100,00 newborns per year, (97% of the approximately 63,000 born per year in Colorado plus roughly 30,000 babies born in military and other government facilities that contract with Colorado for testing and approximately 6,000 babies born in Wyoming each year). Dr. Bill Letson serves as medical director of the program. All newborns are screened by a heel stick blood spot for PKU, biotinidase deficiency, galactosemia, hemoglobinopathies, hypothyroidism, congenital adrenal hyperplasia and cystic fibrosis. These screens are done at the CDPHE laboratory and the results down-loaded to PSD. At PSD, a follow-up case manager is responsible for assuring clinical follow-up for all infants with an initial positive screen for the diseases listed above. Confirmed cases are referred to their primary care providers and the case manager provides referral to an appropriate sub-specialist on a case-by-case basis. No further follow-up by the Newborn Metabolic Screening Program is performed and it is unknown whether the children have a medical home. The state charges \$38.85 for the screen; this funds the laboratory (including a second screen performed at 10 days to two weeks), administration, and follow-up portions of the program. In addition, it partially funds diagnostic testing and treatment in subspecialty clinics, for children diagnosed with the conditions on the screen. It also partially funds the Genetics Counseling Outreach Clinics. All confirmed newborn screens are forwarded to Colorado Responds to Children with Special Needs for inclusion in the birth defects monitoring and prevention program. Newborn Metabolic Screening is guided by an Advisory Committee consisting of physician representatives, a newborn nursing representative, representatives of specialty clinics and two consumer positions (see Attachment 6 for a complete list of members). The Advisory Committee meets quarterly and is responsible for recommending changes in procedure, including which conditions are screened. Recently, the Committee has been investigating the addition of tandem mass spectroscopy. Receiving executive and legislative approval for the new technology has been a barrier to its introduction, despite committee support.

Additional activities include:

- [Folic Acid National Education Campaign](#)
- [Birth defects research](#)
- [Birth defects surveillance](#)

Within CDPHE, the Division of Prevention, Intervention and Treatment Services for Youth and Families (PSD) (genetics services, newborn screening follow-up, Health Care Program for Children with Special Health Care Needs, newborn hearing screening, data information systems, children's and women's health) works closely with the Laboratory and Radiation Services Division (newborn screening laboratory), the Disease Control and Environmental Epidemiology Division (Colorado Responds to Children with Special Needs; Folic Acid Task Force), and the Emergency Medical Services and Prevention Division (Colorado Cancer Registry, and other chronic disease projects).

A specific example of a current collaborative project involving contributing and sharing genetics-relating information, and involving multiple divisions within the health department, is a joint grant to coordinate the various infant, child, maternal, and SIDS mortality reviews administered variously by PSD, the Health Promotion and Disease Prevention Division, and the Center for Health and Environmental Information and Statistics. CDPHE maintains close contact with local health departments through the Public Health Nursing Section in the Office of Local Liaison, which collaborates with public health nurses at the county public health agencies to plan, implement, and evaluate public health programs at the local level. These local public health agencies, in turn, work closely with private physicians in their local communities, thereby bridging the public and private sectors and providing a link and a conduit for information flow in either direction. The local chapters of the March of Dimes and the American Academy of Pediatrics, as well as other public and private agencies and providers participate with CDPHE on many boards and task forces.

The Colorado Consortium of Intensive Care Nurseries (Consortium) also demonstrates many areas of collaboration. Because the Consortium identifies all infants eligible for Part C services in the NICUs based on presumptive eligibility, they perform early referral of those infants identified with genetic diagnoses and hearing screening failures. These referrals assure that infants are identified and referred as early as possible, beginning with an Individual Family Service Plan (IFSP) in the NICU. Public Health Nurses are also involved in each of the Consortium's NICU teams, and receive referrals directly from the NICU. The Public Health Nurses have their own group within the Consortium which meets regularly to problem solve and make sure appropriate identification and referral of infants is made as soon as a diagnosis is acquired. Dr. Joy Browne, Consortium Director, also serves on the advisory committee for Newborn Hearing Screening.

a. Title V Priorities As They Pertain to Genetics-related Services

The Family and Community Health Services divisions of the CDPHE currently provide access to clinical services and counseling for high risk pregnancies and children with metabolic disorders identified through the current newborn screening program. Additionally, funding from Title V of the Social Security Act/Maternal and Child Health Block Grant supports the Health Care Program for Children with Special Health Care Needs. From this location, Genetics activities are integrated with activities that have genetics related components in other Divisions of CDPHE: The Birth Defects Prevention and Monitoring Program (CRCSN) is in the Disease Control and Epidemiology Division, the Chronic Diseases Program in the Health Promotion Disease Prevention Division and the Robert Wood Johnson Turning Point Initiative on Health Disparities in the Executive offices of CDPHE. The Genetics Program has also worked collaboratively with the Environmental Health Division in providing consultation on issues relating to environmental toxicology and teratology.

4) Other Services - Adult

The state provides relatively little provision for specific adult-related genetics services to date. CDPHE does administer the Health Promotion and Disease Prevention Division

Cardiovascular Health through the Chronic Disease Section of the Colorado Department of Public Health and Environment. The program is designed to 1) reduce premature morbidity and mortality from cardiovascular disease, and 2) promote healthy lifestyles for all Coloradoans.

A 10-year cardiovascular disease strategic plan was developed through the Cardiovascular Disease Prevention Coalition, a broad-based coalition that targets the risk factors of 1) high blood pressure, 2) high blood cholesterol, 3) smoking, 4) obesity, and 5) physical inactivity. Specific priority activities identified in the 10-year plan include 1) nutrition, 2) screening, 3) physical activity, and 4) surveillance.

Morbidity, mortality, and cardiovascular disease risk factor data are reviewed on an ongoing basis. The program also coordinates data and educational materials with the National Heart, Lung, and Blood Institute (NHLBI), providing materials on a variety of topics: high blood pressure, high blood cholesterol, smoking, overweight, physical activity, congestive heart failure, asthma, sleep disorder, apnea. The NHLBI Publication Catalogue contains brief descriptions of all their materials and pictures. Pamphlets, posters, reports by national working groups on blood pressure, blood cholesterol, asthma, and other topics can be ordered. Most items are free, and additional materials are available through the American Heart Association of Colorado has numerous print and video resources on a variety of heart disease related topics.

Also providing resources for the public health is the Colorado Comprehensive Cancer Prevention and Control Program (CCPC), which is administered through CDPHE. Funded through the Centers for Disease Control, grants and in-kind donations from coalition partners and other organizations, the program helps coordinate efforts to promote cancer prevention and control activities; and identify and address barriers to appropriate screening, diagnosis, treatment and aftercare.

A fully collaborative program, the CCPC assists with design and implementation of public awareness and education efforts, assists with identification of data sources, recommendations for data analysis, and review and interpretation of data of planning and evaluation. The program also helps assess and contribute to policy development on multiple levels and advocates change to promote comprehensive cancer prevention and control practices. Data sharing through the program also supports a Cancer Registry, which has potential as an information resource in the future regarding genetics-related cancer issues.

5) Indirect Genetics Services – Primary Care Providers and Nurses

The relative paucity of genetics specialists and facilities available to service rural populations places an additional responsibility upon primary care providers, and public and private health nurses.

- There are currently 1,802 Primary Care Physicians practicing in the State of Colorado
- There are approximately 150 Public Health Nurses in Colorado (approximately 125 practicing in rural areas)

The Public Health Nursing Section assures the availability of high quality public health nursing programs in Colorado, which are available through 14 local health departments and 39 county nursing services. The Section also assists local and rural public health agencies in the recruitment and retention of a qualified work force, including public health nurses.

The state health department works with local health agencies to:

- Implement and evaluating public health programs
- Assure that local public health nurses provide safe, competent, legal, and ethical care
- Develop new public health programs to meet the evolving health needs of local communities

Unfortunately, many Primary Care Physicians (PCPs) and Public Health Nurses are generally under informed regarding current medical genetics issues and information.

6) Laboratory Services

Colorado benefits from the availability of excellent clinical laboratory services, which provide a relatively full spectrum of the diagnostic and prognostic test modalities currently available for various inherited and acquired diseases. The state maintains a Public Health Laboratory Division within CDPHE. This lab does testing for communicable diseases, some toxicology and the newborn blood spot screening for the state and outside contractees as noted above. Of the seven private laboratories, two cytogenetics, a biochemical genetics, and a DNA diagnostic laboratory are affiliates of the University of Colorado. The three other facilities are commercially owned and operated. One, Penrose, provides prenatal and cancer diagnostics; Kimball provides molecular testing for prenatal, pediatric and adult disorders; the third, RGC, is focused on reproductive and prenatal genetics.

Colorado Laboratories

Penrose Cytogenetic/Immunopathology

2215 North Cascade Avenue, Colorado Springs, CO 80907
Director: V. Ramesh Babu, PhD
Phone (719) 776-5678; (800) 942-9753
FAX (719) 444-8538

Colorado Genetics Laboratory
University of Colorado School of Medicine
The Children's Hospital - Cytogenetics Laboratory, 4200 East 9th Avenue, #C225, Denver, CO 80262. Director: Loris McGavran, PhD
Phone (303) 315-7249; FAX (303) 315-7044

Kimball Genetics, Inc.
101 University Boulevard, #350, Denver, CO 80206.
Director: Annette Taylor, PhD
Phone (303) 320-1807; FAX (303) 388-9220
Email aktaylor@usa.net

Reproductive Genetics Center (RGC)

Cytogenetics Laboratory, 455 South Hudson Street, Level III, Denver, CO 80246
Directors: George Henry, MD, David Peakman
Phone (303) 399-5393; FAX (303) 399-9160

University of Colorado Health Sciences Center
Department of Pediatrics
4200 East 9th Avenue, #C225
Denver, CO 80262

Biochemical Genetics Laboratory

Director: Steve Goodman, MD
Phone (303) 315-7301; FAX (303) 315-8080

DNA Diagnostic Laboratory

Director: Elaine Spector, PhD
Phone (303) 315-8415; FAX (303) 315-0349
Email Elaine.Spector@uchsc.edu

G) Current Utilization and Perceptions Among Key Medical Providers

1. Overview

Although projections indicate that primary care physicians (PCPs) and public health nurses are likely to be the front line contact for patients needing or receiving genetic services, initial research indicates that neither group is prepared to meet the need to screen, evaluate and refer as necessary based on a patient's symptoms and/or risk factors.

There are multiple contributors to this situation, including limited academic training in genetics in current medical and nursing programs; preponderance of practitioners who were trained before the genetic revolution of the past decade; limited awareness of genetic services; financial pressures from payers within managed care; and limited time to review literature and new developments within subspecialties.

Specifically, two separate studies within the past three years provide insight into medical professionals' preparedness and decision influencers. Highlights of the results of the surveys follow, with the full reports contained in Appendices F and G.

2. Primary Care Providers Survey

Purpose: To document primary care physicians' utilization and perceptions of genetics services.

Methods: A randomized survey of physicians in the Pacific Northwest. Surveys were delivered to 4,824 physicians, including 1,336 internists, 1,227 obstetricians/gynecologist, 1,078 pediatricians, and 1,183 family physicians. Completed surveys were received from 1,642 (34%) respondents, including 401 internists, 394 obstetricians, 436 pediatricians, and 411 family physicians.

Results: The greatest factor prompting a genetics referral was the patient's interest in the evaluation, and the most common reason not to obtain a consultation was the perception that it was of no benefit to the patient. Genetics consultation was rarely sought for a family history of cancer or for deafness, polycystic kidney disease, or congenital heart disease. Even when uncertain about relative risk, physicians usually counseled a patient themselves rather than referring to a specialist.

Physicians asked 59% of essential history items. They frequently obtained appropriate information about presenting symptoms and medications, but they often missed important information about related symptoms and medical history. Physicians frequently screened for smoking and alcohol use, but rarely asked about recreational drug use. Although board-certified general internists performed more comprehensive histories than board-certified family practitioners in the same amount of time, both groups of providers missed a large number of items that should have been influential in developing diagnostic and treatment plans.

Conclusions: Primary care physicians may miss important patient information in their initial interactions with patients. Medical intake questionnaires or other

approaches should be considered to ensure that more complete and accurate information is available to guide diagnostic and treatment plans. Primary care physicians need more education about the genetic component of many diseases to provide directly and to refer appropriately for genetics services.

3. Public Health Nurses Survey

Purpose: To gather background information from Colorado community and public health nurses on the existing level of knowledge in human genetics, current and preferred methods for receiving continuing education, and active interest in having access to continuing education in medical genetics.

Methods: A survey of 102 community and public health nurses was conducted in April 2000.

Results: The survey of public and community health nurses indicated a severe lack of current education and virtually no continuing education options. Selected results included the following:

- 62 % had a college degree
- 80% had some education in genetics while in college
- Nearly 53% of the respondents had been practicing for more than 20 years
- More than 88% cited no continuing education in genetics

The survey also asked respondents to indicate preferred topics for genetics education. Thirty-three percent asked for more information regarding the role of nurses in providing genetics information to patients; 26% asked for information about genetics-related factors relating to high risk pregnancies.

H) PROJECTED NEED FOR SERVICES BY 2005

1) Priority Needs

a) Health Status Indicators

Development and analysis of Public Health Status Indicators has been critical to the development a relevant programs for targeted segments of the population in need of medical services. Although still relevant, the current Health Status Indicators will need to be augmented and updated to generate sufficient data to address population segments and drive service provision and policy development.

b) Redefining “at risk” populations.

According to Dr. Francis Collins of the National Human Genome Research Institute, by the year 2010, screening tests will enable anyone to gauge his or her unique health risks, and genetic discoveries will trigger “a flood” of new pharmaceuticals aimed at the causes of diseases rather than the symptoms.

To date, “at risk” portions of the population have been defined by relatively narrow parameters, including maternal age, household income below poverty level, and ethnicity. However, beyond neonatal and prenatal testing, the genetic screening or diagnosis of relatively common disorders such as certain cancers, cardiovascular disease and mental health disorders in adults could affect a broad segment of the population. Expanding services delivery and reimbursement to cover extended definitions of “at risk” populations will become impossible under the current system.

While predictive timing for these advances is subjective, the reality remains that genetics is rapidly changing the practice of medicine. To date, more than 1900 disorders associated with specific genes have already been identified (see Online Mendelian Inheritance In Man (<http://www.ncbi.nlm.nih.gov/omim>)). By 2005, it is logical to project that additional population screens for hereditary conditions such as hemochromatosis, the most common genetic condition in the United States, and one in which intervention can significantly reduce morbidity, will be in high demand—if not dictated by public policy.

Following rapidly on the heels of increased screening for hereditary disorders are new diagnostics to detect and define risk factors for widespread chronic diseases including cancer, cardiovascular disease, dementia and mental health disorders, diabetes, obesity, blood and immune disorders, and birth defects. For these and other genetics-related health issues, there will be an enormous need to educate the population about the importance of being screened to determine risk factors. It will also be important to implement preventive health programs and strategies to help promote their success in undertaking and (more importantly in the long term) sustaining positive behavioral change.

c) Gaps in Services

The tremendous ramifications of the situation emphasize the current and widening gaps in services to address these needs. In the provision of equitable access, education and new policy development, Public Health will be challenged economically and temporally. The resources in expertise, funding and facilities for genetics related services simply do not exist, nor are they likely to be available quickly, to meet broad public need. Current State genetics programs deal almost exclusively with newborn screening for selected metabolic disorders. Child health programs (see Existing Genetics-related Public Health Programs) are geared towards previously identified disorders which affect relatively small percentages of the population. Additionally, these programs, generally funded by Federal grants, are currently challenged to maintain even their current budgets, and projections of budget cuts loom in the wake of a declining economy.

Additionally, programs which do pertain to adult services need to be updated to encompass aspects of genetics which impact their target populations. Programs such as the Health Promotion and Disease Prevention Division Cardiovascular Health will need to update its risk analysis, correlating in genetic factors. Screening and surveillance

efforts will also need to change as genetic data is added to patient profiles to improve outcomes and long term management.

Other gaps directly impact the potential to reach parity in provision of services throughout the state. For example, although state-of-the-art laboratory services are available, their location is completely concentrated within the major population centers. No localized services are available on the Western Slope, or in the southern region of the state. Because the laboratories are centers for extended consulting and patient follow-up services, the paucity of laboratories in outlying areas affects not only delivery of testing services, but also the critical need for patient information, education and professional medical consultation. As identification and treatment of diseases with a genetic component become a larger part of standard healthcare, this lack of laboratory-centered genetic services will become a greater problem, severely reducing equal access and broad availability of services for the population in rural parts of the state. Health agencies will need to assure the availability and quality of laboratory and clinical services as well as the quality of genetics services, probably in concert with regional or national laboratories. This most likely will require regulatory and statutory measures such as licensing regulations of professional and laboratory services. While cost and quality assurance issues will for the most part preclude establishing new laboratory facilities and counseling locations in rural areas, other remedies, such as centralized (possibly web-based) repositories of genetic testing information, forms and advice, with oversight by certified genetic counselors, could provide a partial solution.

d) Standards of Care in Genetics Services: Current and Projected Models

The Human Genome Project has accelerated the pace of gene discovery leading to the development of an increasing number of genetic tests with broad applications for diagnosing and predicting disease as well as for determining individual response to therapy. In light of this progress, current standards of provision as they apply to genetics testing would appear to require rapid reassessment and evaluation. New criteria to assess the benefits and risks of new genetic tests are mandatory.

e) Genetics Services Evaluation System

[The Secretary's Advisory Committee on Genetic Testing \(SACGT\)](#) and the Association of State and Territorial Health Officers recently recommended enhancing the oversight of new genetic tests to ensure their safety and effectiveness. SACGT determined that a higher level of scrutiny (level II) be considered for tests used for population-based testing or for common disorders (particularly where significant social and ethical issues exist and/or no treatments are available) compared to a lower level of scrutiny (level I) for tests performed for rare diseases or primarily for diagnostic purposes.

Additionally, both groups recommended a series of criteria specifically to determine analytical validity, clinical utility and social consequences in the assessment of the benefits and risks of new tests as they become available. Determinants include the

purpose of the test, whether for prognostic or diagnostic purposes, and possible outcomes. In general, SACGT contends that the greater the uncertainty about the health outcomes associated with a test result, the greater the potential harms of the test. The recommendations take into consideration the effect of positive and negative results for both the patient and family. They include:

- i. Privacy and Confidentiality: The prevention of improper medical disclosure protects individuals from discrimination and serves to strengthen the doctor-patient relationship. In the past, the disclosure of genetic information has led to discrimination and stigmatization. Yet, medical information is essential to conducting genetics and other types of research, and medical records information is particularly important for public health surveillance activities. In order for individuals to feel comfortable in participating in research or testing, it is essential to protect their privacy. Therefore, information resulting from medical services, including genetics, must be treated confidentially and safeguarded from discriminatory misuse.
- ii. Genetic Discrimination in Insurance and Employment: According to studies, fear of insurance or employment discrimination prevents individuals from participating in genetic testing. Information gleaned from genetic tests can diagnose disease, indicate a course of intervention, or provide individuals with information they desire to make life choices. Thus, individuals who avoid testing may miss opportunities to monitor and minimize disease sequelae. Individuals should not be forced to choose between their health and financial security. Therefore, legislation that prevents insurers and employers from discriminating against individuals based on their genetic makeup must be enacted.
- iii. Population-based Screening: State health agencies have been leaders in population genetic screening for more than a quarter century via newborn screening programs. These programs have led to the early diagnosis of mostly rare disorders and have prevented unnecessary morbidity and mortality. As the genetic nature of common diseases becomes more precise, public health will have to address the integration of genetic testing into screening procedures for common late-onset disorders. State health agencies will need to closely monitor the development of genetics tests that can improve screening methods for all common diseases. Furthermore, genetic screening should be accompanied with the appropriate education and counseling, and resources will need to be identified to accomplish this.
- iv. Public Health Workforce Competencies in Genetics: The integration of genetics into public health will heavily depend on the workforce's ability to comprehend genetics information and translate it into existing programs. The CDC has developed competencies based on the multiple disciplines and roles of public health practitioners. These competencies establish a minimum level for genetics knowledge. The state will need to encourage full support and adoption of these competencies by all health agencies.

- v. Eugenics: In the early to mid twentieth century, approximately 30 states enacted eugenics laws to “clean the gene pool” of unwanted characteristics such as mental retardation, leading to the sterilization of tens of thousands of men and women. These programs were thought to be in the public interest. As the integration of genetics into common medical practice accelerates, it is imperative that no programs that infringe upon a person’s bodily integrity or restricts his/her reproductive freedom based on genetic information be allowed to develop.

f) Medical Home Model

Coordinating and improving the services available to children and adults with special health care needs, including children identified with genetics related disorders through newborn metabolic and hearing screening, and those with Cleft Lip/Palate, can be facilitated greatly in the future through the establishment of a Medical Home for these children and their families. A Medical Home model can be developed to facilitate the coordination of services and improve communication between public health and private providers, taking advantage of all possible synergies for the benefit and optimum welfare of the patient.

I) CURRENT DATA COLLECTION SYSTEMS

Data review, data exchange and update of multiple resources for population and genetics services information will be critical to developing and maintaining a current Colorado State Genetics Plan. As more of the population becomes directly affected by the availability of new genetics tests as they are developed, full access to data to confirm relevant population and individual information will be critical. Moreover, issues regarding access to data and privacy will need to be addressed simultaneously, creating additional responsibility on the part of the state to fully define its models for access, provision, education and information management in the delivery of genetics services.

a) Integrated Data Systems

CDPHE has recognized the need to integrate the results of data collection among various state programs in order to best serve the needs of public health. In order to develop highly useful patient health profiles, efforts are underway or being explored to merge multiple state databases, including Children with Special Needs, Newborn Screening, Hearing Screening, Perinatal, Folic Acid, Fetal Alcohol Syndrome, Cancer, Sickle Cell, Maternal, Infant and Child Health Mortality databases. The State's Information Services unit processes and distributes these data as well as Bureau of the Census data and economic data related to population distribution and change. Recent updates in January 2000 to many of these data are available at the Colorado State website Information Services page. All data links to the Office of Genetics and Disease Prevention for the Centers for Disease Control and Prevention.

Currently, the data available through these channels is used by the Maternal Child Health Bureau, U.S. Department of Health and Human Services, Health Resources & Services

Administration, to assess the progress of states in improving the health status of women, infants, children, and adolescents. Planners at the Colorado Department of Public Health and Environment use the information for assessing progress in Colorado, and local organized health departments and nursing services use the data for assessment and planning at the local level.

In addition CDPHE is currently working on an integrated electronic data system as part of the CDC funded Early Hearing Detection and Intervention grants. Colorado's effort is housed in the Children with Special Health Care Needs component of Title V. The primary purpose of this effort is the integration of existing universal newborn metabolic screening (NBMS) and newborn hearing screening (NBHS) data. The databases are to be integrated into an existing electronic platform called the Integrated Registration and Information System (IRIS). What is to be gained from this is:

- 1) *Infant Case Management and Follow-up*: To enhance and assure long term follow-up and case management of infants with specific diagnoses from the NBMS or NBHS and to assist health care providers in follow-up of infants' special conditions and routine health care status.
- 2) *Data for Aggregation and Use in Disease Surveillance*: To establish the template for a Maternal Child Health Disease Surveillance System that will take the form of a Child Health Profile, making it possible to analyze data on a variety of child health issues and use those analyses to guide program and policy from the Colorado Department of Public Health and Environment (CDPHE).
- 3) *Information for Health Care Providers on their Clients*: Create medical provider access to Child Health Profile information for the clients in their medical care, to assure a medical home for the clients and to help establish a "Virtual Medical Home" for the clients within the registry.

b) Access

[The Colorado Health Information Dataset \(CoHID\)](#) provides a queryable format at the state and county level for births, deaths, population, and behavioral risk factors. The database contains the most recent [1999 Colorado birth and death data](#), providing information at the county level in many tables.

[The Colorado Department of Public Health and Environment Statistics and Research](#) home page contains links to other data, including the following, which are fully listed with website links at the end of this document:

- Colorado Registry for Children with Special Needs
- Colorado Central Cancer Registry
- Center for Disease Control, Office of Genetics and Disease Prevention
- Human Genome Epidemiology Network (HuGE Net)
- National Center for Biotechnology Information - Online Mendelian Inheritance In Man
- Mountain States Genetics Network
- GeneTests - a directory of clinical laboratories providing testing for genetic disorders

- Colorado Department of Local Affairs
- CORN *Guidelines for Clinical Genetic Services for the Public's Health*

c) Implications

For the purposes of developing and implementing a Colorado State Genetics Program, these sources of available data will continue to be invaluable in the creation of population health profiles. By correlating geographic, economic and racial/ethnic data, programs can direct support to areas of need and thereby improve outcomes.

Coordination of these data also is fundamental to the development of state policy. Cooperation between state and federal public health programs to share program data becomes more critical as genetics becomes a larger part of core public health functions. Population-based data and measurement of health outcomes will help set new standards for core programs, and the achievement of improved public health through the use of genetics for early diagnosis and efficient patient management are likely to greatly impact how existing core programs evolve. Much of the information from Title V programs is already analyzed to identify disparities and gaps in services among different segments of the population.

J) Needs Assessment Conclusions

Given the plethora of data from both scientific and popular sources, it would be naïve to assume that there is not a need for an evolving, comprehensive Colorado State Genetics Plan to serve current and future population needs. Technological advances continue to negate arguments regarding the provision of services for “special needs populations”, since the ubiquity of the human genome and its implications in all aspects of health are indisputable. Education and equitable services delivery to an informed population must now be considered compulsory.

The challenge has become one of time and appropriateness, coupled with the inevitable issues surrounding the economics and ethics of expanded genetics services provision. Scaling a relevant program to fairly meet the needs of the target populations at various points in time appears to be the seminal focus of the first iteration of any state genetics services program. As the program is developed over the next few months, the collaboration of existing state services, special interest groups, medical and scientific professionals, policy makers and influencers, and especially consumers, will be critical factor in ensuring the development of a workable services delivery platform.

V. State Genetics Plan Development and Implementation Recommendations

A) Mission, Goals and Objectives

Emerging genetics information is widely expected to have far-reaching impact in public health. A major consequence of the Human Genome Project and other genetics research has been the mounting evidence that birth defects and chronic diseases such as cancer, cardiovascular disease, dementia and mental health disorders, diabetes, obesity, blood and immune disorders all have major, identifiable genetic risk factors that can act in concert with personal behaviors and other environmental risk factors to cause disease in virtually all segments of the population. There is a pressing need to use the information to develop public health interventions that will meet the following mission and goals:

Mission – To realize fully the goals of Healthy People 2010 through the seamless integration of quality genetics services into health care systems in Colorado.

Goals – Long Term

To improve outcomes, and when possible prevent morbidity and mortality among Coloradans with, or at risk of, genetic, congenital and/or hereditary disorders by assuring the continued review, assessment, integration and delivery of genetic services to all individuals and families who may be at risk. This will include addressing various underserved segments of the population, including low income and ethnic groups, women and seniors currently unaware of, or without access to, evolving genetics-based health care services. Meeting long term goals will eventually involve developing protocols for the creation of inclusive programs to routinely update existing public and private health care systems with validated genetics-related medical services.

Goals – Short Term

1. Ensure the effective and efficient development, implementation and review of the State Genetics Plan by re-establishing the State Genetics Coordinator position. As a full-time position (FTE), it will be the role of the State Genetics Coordinator to coordinate activities of the Genetics Advisory Committee, to facilitate policy development, coordinate communications, services integration and collaboration between public health and private services, and coordinate on-going program evaluation and quality improvement processes. This role is a critical factor in the efficient development and implementation of the State Genetics Plan.
2. Assure a collaborative, culturally diverse program that links with community partners and addresses health disparities and underserved populations. This goal will be supported by the continued maintenance of the State Genetics Advisory Committee (GAC), which will help develop an infrastructure for the planning, implementation, monitoring and evaluation of genetics services in Colorado.
3. Support the development and implementation of the Medical Home system, providing a virtual Medical Home for patient information and eventually a complete electronic medical record for case management purposes as described in the current Maternal Child Health Bureau Grant application.

Objectives

1. Support the development and implementation of an expanded Newborn Metabolic Screening program using Tandem Mass Spectrometry (MS/MS) to increase identification of additional metabolic disorders which are pre-symptomatically treatable. This program would provide a model for collaborative development and provision of genetics-related healthcare services through public health services, university-based services and private providers.
2. Support the development and implementation of an Adult Medical Genetics Program at the University of Colorado Hospital (UCH). Like the Tandem Mass Screening program, this program would also provide a model for collaborative development and provision of shared genetics-related healthcare data and services through public health services, university-based services and private providers.
3. Develop evolving education programs to create equal population awareness regarding genetic services among consumers, including underserved populations and ethnic groups, medical health care providers, policy makers, legal professionals, insurance providers, clergy, the media, teachers and students.

B) Colorado State Genetics Plan – Program Matrix (In process. See attached.)

Organized by target age groups, the State Genetics Plan Program Matrix addresses genetic services currently available, gaps in services, priority issues for the next 3-5 years, recommended strategies to address these priorities, the state's role, identification of other stakeholders as it pertains to service priorities, and action items and timelines. As all aspects of the program are currently evolving, the matrix will similarly evolve, with course corrections, new opportunities and stakeholder input from the public health and private practitioner perspectives.

C) Action Items - Program Development and Coordination

1. Oversight and Coordination of a General Policy Development Model

a. Action 1 - Re-establishment of the State Genetics Coordinator

The State Genetics Coordinator will play a pivotal role in bringing collaborative partners together to work toward accomplishing State Genetics Plan objectives. Due to the required collaboration between public health and private resources to service adequately serve all segments of the population, including low income families, ethnic groups, seniors and other underserved individuals, the State's and public health's interests can only be guaranteed through oversight from this position.

The State Genetics Coordinator position was originally funded as part of the MCH grant, and defined by the CORN Guidelines for Clinical Genetics Services for the Public Health. That funding ended at the close of FY 2001.

b. Action 2 – Maintenance of the Genetics Advisory Committee

Established in 2000 as part of the implementation of the genetics portion of the Maternal and Child Health Block Grant, the Genetics Advisory Committee (GAC) is comprised of a cross section of public health professionals, private health care professionals from the commercial and non-profit sectors, policy development experts, ethicists, legal and legislative professionals, clinical services providers, educators and consumers. The Committee participates in an ongoing dialogue on legal, social or ethical issues arising from genetic research with emphasis on how these issues impact various Colorado publics. Consistent with support of the State Genetics Plan, the GAC examines, quantifies and extrapolates the growth of genetics services based on developments in medical applications in the past two- and five-year periods. With the guidance of the State Genetics Coordinator, the GAC reviews existing, secondary sources and channels of information regarding genetics and genetics services to assess specific areas of need, correlate with the existing services availability, extrapolate need based on the projected growth of medical genetics services, and project appropriate expansion of services and resources over the next five years.

In conjunction with the State Genetics Coordinator, the GAC will analyze all data in review of the State Genetics Services Delivery Plan that will provide practical guidelines and resources from both the State's and the population's perspective. Additionally the group will issue a report on the barriers to receipt of appropriate genetic services in underserved areas or populations, along with recommendations to the state legislature/executive branch on policy direction and legislation.

The GAC has also adopted an extended agenda which includes additional assurance and policy development activities such as presentations to primary care providers in key forums to inform them of the impact of genetic research on patient education; meetings with legislators to inform them on possible policy directions and/or proposed model legislation; and coordination of consumer public forums around the state regarding genetic illnesses or conditions, prenatal diagnosis, and current diagnosis or treatment options.

D) Action Items - Service Provision Options, Collaborations and Coordination

Partnerships between public and private health care providers are essential to the success of this project. Collaboration between existing State public health programs, university-based programs, private practitioners, health maintenance organizations (HMOs) and other health care resources already exist in the provision of some genetics-related services. As additional model programs evolve, the State Genetics Coordinator will be act as liaison between these groups to facilitate collaboration. Initial models, such as the Medical Home program and the Tandem Mass Spectrometry programs to extend newborn metabolic

screening (described below), will provide examples and opportunities to improve interaction and enhance services. Additionally, a proposed Adult Medical Genetics Program through the University of Colorado Hospital offers a unique opportunity for cooperation and collaboration between university-based programs, state health department resources and other health care providers involved with patient follow-up services.

1. Action 3 - Medical Home

One of the national Healthy People 2010 objectives promoted by the Maternal and Child Health Bureau is to assure all children with special health care needs will receive ongoing comprehensive care within a medical home. A medical home is defined as a regular source of primary health care that is family-centered, accessible, community-based, coordinated, compassionate, and culturally respectful. To address this objective, the Health Care Program for Children with Special Needs (HCP) has created a Medical Home Initiative in Colorado involving a broad and diverse group of stakeholders who serve as advisors and/or participants in workgroups.

The Medical Home Initiative supports six objectives that will enhance the development of a medical home for those children identified through Newborn Metabolic Screening and Newborn Hearing Screening programs. They Are: 1) Development of a Medical Home Resource Library; 2) Development of a Model, Family-Directed Health Record; 3) Heightened Awareness of Medical Home Across Stakeholders; 4) Development of a Medical Home “Clinical Laboratory”; 5) Medical Home Care Coordination; 6) Data Collection and Evaluation. The data collection and evaluation efforts have produced disturbing facts indicating that many children in Colorado do not have access to primary health care. Approximately 10% of children are uninsured and are unlikely to be receiving primary care. Approximately 20% of children are enrolled in publicly funded health insurance programs, Medicaid and CHP+, but the capacity of participating primary care providers (PCP) and the recent dramatic decline in participating providers creates a new and alarming access issue. This issue will be a priority of the Medical Home Initiative.

The complexity of diagnosis, treatment and follow-up for the growing list of genetics related disorders requires a highly collaborative effort to provide access to pertinent medical information and family health history. The development of a virtual Medical Home will provide integration of public health services through system linkages and genetic program resources to ensure the needs of the population are met. As addressed in the current Maternal Child Health Bureau SPRANS Grant application entitled “Genetic Services-Improving Health of Children: Implementation of the State Grants for the integration of Programs and Their information Systems”, the Medical Home data storage system will instigate collaboration and continuity of health care services, particularly between public health providers and private medical

provider groups. Such collaboration, which has already been endorsed by the Colorado Chapter of the American Academy of Pediatrics and the Colorado Academy of Family Practice.

2. Action 4 – Collaboration and Support of University-based Services

Developing ways to coordinate existing resources for the welfare of public health currently include collaboration with genetics services available through the University of Colorado Health Sciences Center and the University of Colorado Hospital (UCH). These entities provide referral resources to medical genetics specialists, genetic counseling and laboratory services.

To extend the limited genetics resources currently available to adults, the State Genetics Plan supports the development and implementation of the proposed Adult Medical Genetics Program at the University of Colorado Hospital. Extending adult services beyond the established cardiovascular genetics clinic, the program would initiate a general genetics clinic and would serve as models for developing future clinics that can target other genetics-related disorders in individuals and families. This program will address an important and underserved aspect of adult care. Like the Tandem Mass Screening program, this program would also provide a model for collaborative development and provision of shared genetics-related healthcare data and services through public health services, university-based services and private providers.

3. Action 5 - Private Providers

Due to limited resources and economic constraints, the state's role in public health is defined as one of assessment, coordination, assurance, policy development, and education. This is particularly true as it pertains to genetics and public health. As genetics impacts new areas of health care, large numbers of the population will be affected, to the point where it will eventually be desirable and practicable to develop patient health profiles and possible options for services for all individuals.

Under these circumstances, the synergies and opportunities for collaboration between public health entities and the private sector are enormous and must be pursued. Currently, managed care programs work with other private providers, university-based programs and the state to deliver minimal genetics-based services. At Kaiser Permanente, for example, children and families identified with genetics-related disorders as part of state-mandated newborn screens are referred to Children's Hospital. Older children, adolescents and adults receive initial physical work-ups through Kaiser and generally referred to University programs if diagnosed. Kaiser also provides on-staff genetic counseling in Denver, some patient education materials regarding preconception and cancer, and has a cancer epidemiology program that interacts with the Center for Disease Control (CDC).

E) Action 6 – Collaboration in the Development of New Program Models and Tandem Mass Spectrometry

In the adoption of new genetic services for the Colorado population, viable models need to be developed through collaboration to show utility, benefit and service delivery to the population on an equitable basis.

The State Genetics Plan will support the development of this type of model utilizing Tandem Mass Spectrometry (TMS). TMS is a new and evolving technology that is being applied to newborn screening services. More than 20 disorders of body chemistry can be detected in a single 1/8-inch dried blood spot.

Beginning in March 2002, there will be limited screening by Tandem Mass Spectrometry offered to infants born in Littleton (a southwestern suburb of Denver) hospitals by a laboratory at the University of Colorado Health Sciences Center (UCHSC). This effort will begin with a maximum of approximately 250 births a month and will likely increase over time. The number and type of disorders detected will depend on the number screened in a given year. The Laboratory and Radiation Services Division, Newborn Screening Laboratory will provide the testing. This is quite wrong! The testing will be done by Goodman's lab at UCHSC. At the moment LARS has nothing to do with this and can't because we've never received permission from the CDPHE and state powers that be. The Prevention and Intervention Services for Children and Youth Division at CDPHE will work with UCHSC on how to provide follow up services.

The TMS program provides an excellent model for evaluation of expanded genetic services and the potential challenges that may arise. For example, the majority of children identified by metabolic screening live in the Denver metropolitan area, but, depending on the disease entity, 20-40% live in rural or semi-rural areas. Follow-up of children living in the Denver metro area by appropriate pediatric sub-specialists is felt to be better than that for children born in rural areas, but neither is likely to be as good as it could be. In terms of long term follow-up for the special medical needs of these children in general, the same urban/rural disparity exists, i.e. the extent of proper follow-up outside the Denver metro area for several of these conditions is uncertain. This is most apparent for congenital hypothyroidism. In this instance, since primary providers and families tend not to feel a need for frequent sub-specialty consultation for children with hypothyroidism, degree of appropriate follow-up and case-management even in the Denver metro area is quite uncertain. The degree to which children diagnosed with conditions identified initially by the newborn metabolic screen have an appropriate medical home, especially one that is solidly in the case-management and referral loop, is largely unknown.

Of additional importance in the development of this expanded screening program is the opportunity to coordinate and collaborate with private providers and managed care entities. Within the next six months, Kaiser Permanente is planning to add expanded newborn screening to its program, using the UCHSC laboratory under the direction of Steve Goodman, M.D. Kaiser will provide follow-up and referral services, and will track patient outcomes, providing important data for program analysis which can be utilized by other entities for evaluation of other extended genetic screening programs.

F) Action 7 – Education

Virtually all areas of review, including public health, medical professionals, consumers and various stakeholders including policy developers, legal support and clergy, are in need of education in existing and emerging genetics services and their impact on public health. As a primary resource for education and information regarding public health issues, the State needs to coordinate a comprehensive education program which will integrate the following components in collaboration with other stakeholders. These programs will be coordinated by the State Genetics Coordinator, and may change in priority as new services and special needs arise among the population.

Priority actions:

Secure a full-time educator (preferably a board certified genetic counselor) for the Newborn Screening (hearing and metabolic testing) programs. This position would provide cross-over education options for professionals and patients in various situations.

1. Collaborate with other public and private entities to co-sponsor educational programs and materials with UCHSC, Children’s Hospital, private providers such as Kaiser Permanente, etc.
2. Develop relationships with insurers to provide education on reimbursement issues, i.e. out-of-state lab requirements, genetic counseling, etc.
3. Extend relationships with medical, nursing, public health, allied health schools and private entities such as the Mountain States Genetics Foundation (MSGF) to integrate genetics into the curriculum as required courses; and to provide continuing education credits in genetics for current practitioners.
4. In conjunction with the Genetics Advisory Committee, develop a Genetic Speaker’s Bureau for all levels of education, from physicians and nursing professionals to secondary education.
5. Develop and distribute a statewide resource directory on CD-ROM and in hard copy of clinical services and indications for referral for health care providers. This directory can be provided in conjunction with the MSGF and the resource section of their website.
6. Optimize links from the State website to other genetics-related resources on the Web.
7. Develop a brochure regarding the benefit and indications for clinical genetic services throughout the lifecycle to assist in educating insurers and policy makers.
8. Expand distribution of the existing resource guides and educational materials available from the former Mountain States Genetics Network. Update as needed through the MSGF.

These materials, and others to be developed, will address the following subsections of the target populations:

- 1) Practitioner Awareness

- a. Primary Care Providers
- b. Public Health Nurses
- c. Private Health Nurses
- d. Population Awareness
 - 1. Preventive Services
 - 2. Maternal and Child Health
 - 3. Newborn Screening
 - 4. Developmental Health
 - 5. Adult Services
 - 6. Health Promotion
 - a. Preconception Education
 - b. Adult Predisposition Awareness
- e. Cross-Audience Education
 - 1. Target Audience Segmentation
 - a. Adoption Workers
 - b. Affected Families
 - c. Allied Healthcare Professionals
 - d. Clergy
 - e. Clinical Genetics Professionals
 - f. General Public
 - g. Insurance Providers
 - h. Law and Policy Makers
 - i. Legal Professionals
 - j. Medical and Professional Association Leaders
 - k. News Media
 - l. Researchers and Institutional Review Boards
 - m. Support Groups
 - n. Teachers

VI. Conclusions

In order to fully realize the potential of the genomic revolution in medicine, a comprehensive program of communication and collaboration between public health entities, private practitioners, patients and patient influencers is imperative. By creating workable models to share data, address gaps in genetic services, special needs groups, education and culturally sensitive issues, a comprehensive state genetics program will evolve that provides equal access to all members of the population.

VII. Appendices (See following attachments.)

- A. Genetics-related Public Health Resources in the State of Colorado
- B. Colorado Department of Local Affairs. *Colorado State Population Projections, 1990-2005*. June 2000. <http://www.dola.state.co.us/demog/project.htm>
- C. *Key Colorado Vital Statistics for 1998*. Demography section, Colorado Division of Local Government. <http://www.cdphe.state.co.us/hs/summaryweb98.pdf>
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- E. *Public and Community Health Nurses Survey, 2000*. Colorado Department of Public Health & Environment.
- F. Current Listing of Hereditary Diseases

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Human Genome Epidemiology Network (HuGE Net)
<http://www.cdc.gov/genetics/hugenet/whatsnew.htm>

Online Mendelian Inheritance In Man (<http://www.ncbi.nlm.nih.gov/omim>), a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere; developed for the World Wide Web by [NCBI](#), the National Center for Biotechnology Information. The database contains textual information, pictures, reference information, links to NCBI’s [Entrez](#) database of MEDLINE articles and sequence information, and the OMIM Morbid Map, a catalog of genetic diseases and their cytogenetics map locations arranged alphabetically by disease.

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Appendix A

Genetics-related Public Health Resources in the State of Colorado

Genetics-related Public Health Resources in the State of Colorado

The Children's Hospital Family Resource Center

1056 East 19th Avenue
Denver, CO 80218
Phone (303) 861-6400

Genetics Services

Penrose Cancer Center Hereditary Cancer Service

103 East Monroe St.
Colorado Springs, CO 80933
Contact: Jeff Shaw, M.S.
Phone (719) 776-5274
Fax (719) 776-2516

The Children's Hospital Specialty Clinics

Cleft Palate Clinic

Phone (303) 837-2574
FAX (303) 861-3992

Cystic Fibrosis Clinic

Phone (303) 837-2522
FAX (303) 837-2924

Fragile X Treatment and Research Center

Phone (303) 837-2598
FAX (303) 764-8086

Inherited Metabolic Diseases Clinic

Phone (303) 861-6847
FAX (303) 764-8024

Muscle Clinic

Phone (303) 861-6633
FAX (303) 861-6066

Neurocutaneous Clinic

Phone (303) 861-6947
FAX (303) 861-3921

Spina Bifida Clinic

Phone (303) 861-6633
FAX (303) 861-6066

Regional Genetics Clinics

Phone (303) 861-6395

FAX (303) 861-3921

Clinic Sites: Colorado Springs, Durango, Grand Junction, Greeley, and Pueblo

University of Colorado Health Sciences Center

4200 East 9th Avenue

Denver, CO 80262

UCHSC Specialty Clinics

Hemophilia Center

Phone (303) 724-0724

FAX (303) 724-0947

Hereditary Cancer Clinic

Phone (303) 372-9113

FAX (303) 372-9129

Huntington Disease

Predictive Testing Program

Phone (303) 315-3601

FAX (303) 315-5467

Colorado Neurological Institute

Huntington Disease Center of Excellence

701 East Hampden Avenue, #530

Englewood, CO 80110

Phone (303) 321-5503

FAX (303) 788-8854

Newborn Screening Follow-up

CLINICAL:

Colorado Department of Public Health and Environment

Medical Consultation and Genetic Services

4300 Cherry Creek Drive South

Denver, CO 80246

Phone (303) 692-2425

FAX (303) 782-5576

LABORATORY:

Colorado Department of Public Health and Environment

Laboratory & Radiation Services Division

Newborn Screening Laboratory

8100 Lowry Boulevard

Denver, CO 80220

Phone (303) 692-3488

FAX (303) 344-9989

Prenatal Genetics Services

Genetic Counseling Services

14475 West 54th Avenue
Arvada, CO 80002
Phone (303) 216-2505

Memorial Hospital

Department of Perinatology

1400 East Boulder
Colorado Springs, CO 80909
Phone (719) 365-5960
(888) 600-2961
FAX (719) 365-5977

Genetics & Women's Health Specialists

3205 North Academy Boulevard
Colorado Springs, CO 80917
Phone (719) 776-3470
FAX (719) 776-3154

Denver Health Medical Center

OB-GYN Department

777 Bannock Street
Denver, CO 80204
Phone (303) 436-6580
FAX (303) 436-7081

Kaiser Permanente

Department of Perinatology and Genetics

2045 Franklin Street
Denver, CO 80205
Phone (303) 839-7341
FAX (303) 839-7360

Presbyterian/St. Lukes Medical Center

Maternal/Fetal Medicine

1719 East 19th Avenue
Denver, CO 80218
Phone (303) 839-7341
FAX (303) 839-7360

Reproductive Genetics Center

455 Hudson Street, Level III
Denver, CO 80246
Phone (303) 399-5393
(800) 399-5577
FAX (303) 399-9160

St. Joseph Hospital
Mountain States Maternal/Fetal Medicine
Midtown 1, Suite 750
Denver, CO 80205
Phone (303) 837-7885
(800) 252-0399
FAX (303) 837-7967

University of Colorado Health Sciences Center
Prenatal Diagnosis and Genetics Center
4200 East 9th Avenue, #E197
Denver, CO 80262
Phone (303) 372-1026
FAX (303) 372-1816

Laboratory Services

Penrose-St. Francis Health Services
Cytogenetic/Immunopathology
2215 North Cascade Avenue
Colorado Springs, CO 80907
Phone (719) 776-5678
(800) 942-9753
FAX (719) 444-8538

Colorado Genetics Laboratory
University of Colorado School of Medicine
The Children's Hospital
Cytogenetics Laboratory
4200 East 9th Avenue, #C225
Denver, CO 80262
Phone (303) 315-7249
FAX (303) 315-7044

Kimball Genetics, Inc.
101 University Boulevard, #350
Denver, CO 80206
Phone (303) 320-1807
FAX (303) 388-9220

Reproductive Genetics Center
Cytogenetics Laboratory
455 South Hudson Street, Level III
Denver, CO 80246
Phone (303) 399-5393
FAX (303) 399-9160

University of Colorado Health Sciences Center
Department of Pediatrics
4200 East 9th Avenue, #C225
Denver, CO 80262

Biochemical Genetics Laboratory
Phone (303) 315-7301
FAX (303) 315-8080

DNA Diagnostic Laboratory
Phone (303) 315-8415
FAX (303) 315-0349

Appendix B

Colorado Department of Local Affairs Colorado State Population Projections, 1990-2005 June 2000

**PRELIMINARY POPULATION PROJECTIONS FOR COLORADO COUNTIES
1995 – 2005**

COUNTY	1995	2000	2005
COLORADO TOTAL	3,782,175	4,250,110	4,653,254
Adams	299,775	336,909	380,954
Alamosa	14,941	16,696	18,258
Arapahoe	446,200	497,310	527,050
Archuleta	7,113	10,043	12,530
Baca	4,443	4,562	4,649
Bent	5,676	6,258	6,501
Boulder	256,737	286,460	309,443
Chaffee	14,868	16,818	18,320
Cheyenne	2,344	2,395	2,423
Clear Creek	8,675	9,331	10,282
Conejos	7,750	8,099	8,373
Costilla	3,411	3,507	3,729
Crowley	4,499	4,938	5,046
Custer	2,718	3,760	4,617
Delta	25,175	28,017	31,803
Denver	500,541	541,835	556,821
Dolores	1,598	1,935	2,103
Douglas	104,623	175,938	225,303
Eagle	28,860	36,513	43,127
Elbert	14,631	20,948	26,727
El Paso	466,172	512,829	557,941
Fremont	40,202	45,055	47,715
Garfield	35,980	43,188	49,533
Gilpin	3,683	4,872	5,285
Grand	9,219	10,892	12,793
Gunnison	11,906	13,913	15,535
Hinsdale	642	755	861
Huerfano	7,071	7,660	8,925
Jackson	1,733	1,834	1,972
Jefferson	491,089	527,790	550,652

Kiowa	1,726	1,787	1,825
Kit Carson	7,309	7,820	8,006
Lake	7,430	8,450	10,536
La Plata	39,190	43,217	49,220
Larimer	217,127	243,414	269,919
Las Animas	15,440	16,322	20,300
Lincoln	6,300	6,705	7,028
Logan	18,527	18,849	22,651
Mesa	106,035	118,211	131,527
Mineral	630	742	822
Moffat	12,111	13,053	13,965
Montezuma	21,965	23,509	26,329
Montrose	29,494	32,871	37,098
Morgan	25,396	27,377	29,840
Otero	21,190	21,292	22,220
Ouray	3,046	3,701	4,450
Park	10,713	15,676	24,953
Phillips	4,536	4,631	4,781
Pitkin	14,372	14,375	16,249
Prowers	13,685	14,220	14,786
Pueblo	130,181	140,786	150,614
Rio Blanco	6,966	7,120	7,889
Rio Grande	11,748	12,347	13,128
Routt	16,718	18,648	21,234
Saguache	5,555	6,311	6,793
San Juan	565	541	611
San Miguel	5,227	6,159	6,925
Sedgwick	2,648	2,755	2,843
Summit	17,248	21,169	25,609
Teller	17,505	22,012	25,712
Washington	5,363	5,205	5,352
Weld	154,519	179,965	210,501
Yuma	9,435	9,810	10,267

Appendix C

Colorado Vital Statistics Summary by County and Region Colorado Residents, Annual Average, 1996-2000

Appendix D

Public and Community Health Nurses Survey, 2000 Colorado Department of Public Health & Environment

Appendix E

Current Listing of Hereditary Diseases University of Washington, Seattle

Current Listing of Hereditary Diseases

22q11 Deletion Syndrome

Achondroplasia

Adrenoleukodystrophy (X-Linked)

Alagille Syndrome

Alpha-Thalassemia X-Linked Mental Retardation Syndrome (also XLMR Hypotonic Facies Syndrome)

Alzheimer Disease Overview

Alzheimer Disease, Early-Onset Familial

Androgen Insensitivity Syndrome

Angelman Syndrome

Ataxia Overview, Hereditary

Ataxia-Telangiectasia

Becker Muscular Dystrophy (also The Dystrophinopathies)

Beckwith-Wiedemann Syndrome

Beta-Thalassemia

Biotinidase Deficiency

Branchiootorenal Syndrome

BRCA1 and BRCA2 Hereditary Breast/Ovarian Cancer

Breast Cancer Genetics - An Overview

CADASIL

Canavan Disease

Charcot-Marie-Tooth Overview

Charcot-Marie-Tooth Type 1

Charcot-Marie-Tooth Type 2

Charcot-Marie-Tooth Type 4

Charcot-Marie-Tooth Type X

Cockayne Syndrome

Contractural Arachnodactyly, Congenital

Craniosynostosis (FGFR-Related)

Deafness and Hereditary Hearing Loss Overview

DRPLA (Dentatorubral-Pallidoluysian Atrophy)

DiGeorge Syndrome (also 22q11 Deletion Syndrome)

Duchenne Muscular Dystrophy (also The Dystrophinopathies)

Dystonia, Early-Onset Primary (DYT1)

Dystrophinopathies, The

Ehlers-Danlos Syndrome, Kyphoscoliotic Form

Ehlers-Danlos Syndrome, Vascular Type

Epidermolysis Bullosa Simplex

Facioscapulohumeral Muscular Dystrophy
Factor V Leiden Thrombophilia
Familial Adenomatous Polyposis (FAP)
Familial Mediterranean Fever
Fragile X Syndrome
Friedreich Ataxia
Frontotemporal Dementia with Parkinsonism-17

Galactosemia
GM2 Gangliosidosis (also Tay-Sachs)
Gaucher Disease

Hemochromatosis, Hereditary
Hemophilia A
Hemophilia B
Hemorrhagic Telangiectasia, Hereditary
Hereditary Hearing Loss and Deafness, Nonsyndromic, DFNA3 (Connexin 26)
Hereditary Hearing Loss and Deafness, Nonsyndromic, DFNB1 (Connexin 26)
Hermansky-Pudlak Syndrome
Huntington Disease
Hypochondroplasia

Ichthyosis, Congenital, Autosomal Recessive
Incontinentia Pigmenti

Kennedy Disease (also Spinal and Bulbar Muscular Atrophy)
Krabbe Disease

Leber Hereditary Optic Neuropathy
Lesch-Nyhan Syndrome
Li-Fraumeni Syndrome
Limb-Girdle Muscular Dystrophy Overview
Lipoprotein Lipase Deficiency, Familial
Lissencephaly Overview

Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
Multiple Endocrine Neoplasia Type 2
Multiple Exostoses, Hereditary
Muscular Dystrophy, Congenital Overview
Myotonic Dystrophy

Nephrogenic Diabetes Insipidus
Neurofibromatosis 1
Neurofibromatosis 2
Neuropathy with Liability to Pressure Palsies, Hereditary

Niemann-Pick Disease Type C
Nijmegen Breakage Syndrome
Norrie Disease

Oculocutaneous Albinism Type 1

Pallister-Hall Syndrome
Pelizaeus-Merzbacher Disease
Pendred Syndrome
Phenylalanine Hydroxylase Deficiency
Prader-Willi Syndrome
PROP1-Related Combined Pituitary Hormone Deficiency (CPHD)

Retinitis Pigmentosa Overview
Retinoblastoma
Rothmund-Thomson Syndrome

Smith-Lemli-Opitz Syndrome
Spastic Paraplegia Overview, Hereditary
Spinal and Bulbar Muscular Atrophy (also Kennedy Disease)
Spinal Muscular Atrophy
Spinocerebellar Ataxia Type 1
Spinocerebellar Ataxia Type 2
Spinocerebellar Ataxia Type 3
Spinocerebellar Ataxia Type 6
Spinocerebellar Ataxia Type 7
Stickler Syndrome (Hereditary Arthroophthalmopathy)

Tay-Sachs (also GM2 Gangliosidosis)
Tuberous Sclerosis Complex

Usher Syndrome Type I
Usher Syndrome Type II

Velocardiofacial Syndrome (also 22q11 Deletion Syndrome)
Von Hippel-Lindau Syndrome

Williams Syndrome
Wilson Disease

X-Linked Dilated Cardiomyopathy (also The Dystrophinopathies)
X-Linked Hypotonic Facies Mental Retardation Syndrome (also Alpha
Thalassemia X-Linked Mental Retardation Syndrome)