COLORADO STATE GENETICS SERVICES PLAN

Colorado Department of Public Health and Environment
Children and Youth with Special Health Care Needs Section
4300 Cherry Creek Drive South
Denver, CO 80246

February 18, 2002
(Revised June 15, 2004)

TABLE OF CONTENTS

I. ACKNOWLEDGEMENTS

The State Genetics Advisory Committee

The development of an integrating genetics plan 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) has assembled a State Genetics Advisory Committee (GAC), consisting of a diverse group of medical practitioners, public health professionals, 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 serve voluntarily on the GAC in two-year, renewable terms. The State Planning Grant provides administrative support to the committee.

Federal and Statewide Agencies and Boards

True to the need for cooperation between various federal and state agencies in the development of public health programs, multiple human and health services agencies interact with CDPHE to create efficiencies through collaboration as new programs are brought to the public. The existing and evolving genetics programs that form the foundation for this report draw upon valuable 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 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 bring into focus 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 conditions caused by 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 these mutations in at risk individuals will lead to better education of how they are passed to offspring and improve informed reproductive decision making.

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 that some gene mutations increase the likelihood of unhealthy living. Moreover, 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 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 providing 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.

Genomics has previously focused on specific disorders affecting a relatively small percentage of the population, but advances in the field have continued 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. Utilize an age-based approach, that is, to define goals according to target demographics including preconception, newborn, young children, adolescents, adults and seniors.
2. Review genetic services currently available to these demographic groups.
3. Determine current gaps in genetics services to these demographic groups.
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. Identify 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 provide state policy makers 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

Background

The challenges facing public health at the dawn of the 21st century reflect a daunting combination of opportunities, ethical ambiguities, and economic realities. While the fundamental mission of public health is to "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 the 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 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 that are relatively rare. A large number of these tests are used in maternal health and newborn screening programs. 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 300 individuals of Northern European descent, are rapidly being developed. 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.

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

Although it is assumed that genetics will play an ever increasing role in nearly every aspect of health care, current provision of services is limited, and the timeframe for the broad application and adoption of genetic medicine is undetermined. 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 to 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 responsibilities must CDPHE then take on for potentially increasing demand for services that are too expensive and too specialized for the general population to access?
- As genetics drives new standards of care, how will new tests and therapies be funded?
- 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?

What is the Responsibility of Public Health?

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. population frequency of genetic variants that predispose people to specific diseases, both common and rare;
2. population frequency of morbidity and mortality associated with such diseases; and
3. 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

2 Koury et al, 2000.

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, and the rapid aging of any needs assessment will be mirrored in lagging services and underserved publics.

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.

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 considered when developing 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. The policy advisements are to:

- 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.

Assurance

Public health agencies assure that their constituents have necessary services to achieve the goals that are provided, either by encouraging action by other entities (private or public sector), or by requiring such action through regulation, or by providing services directly (IOM, 1988). Agencies may collaborate with other public and private entities to educate public 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 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.

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 and in order to create the basis for a state genetics plan for Colorado, several specific areas of focus 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

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.

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 formally beginning to develop 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.

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

Arizona's plan was 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:

- 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 written 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 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.

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; pre-implantation diagnoses will be more widely available; and primary care providers will practice, to some extent, 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 service 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.

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.

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 Public Health and Environment, 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 Public Health and Environment and Human Services.

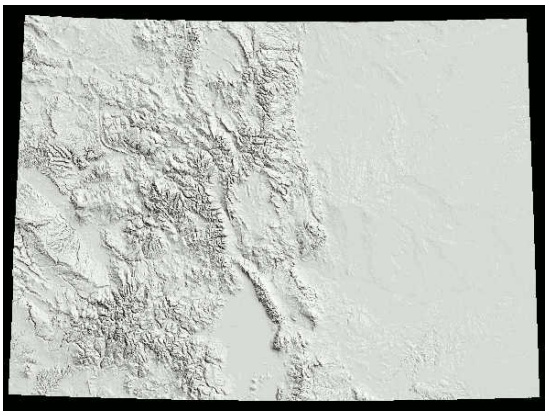
- Recent discussions of genetics in professional and consumer media (newspapers, magazines (see *Bibliography* below).
- Current genetic testing availability and projections for new testing.
- Numbers of primary care physicians and public health nurses currently servicing the Colorado population.
- Existing genetics-related public health systems
    i. Existing genetics services resources
    ii. Projected need for services by 2005
    iii. Reviews
- List of current resources (Public and Private) per the Mountain States Genetics Network and the national *GeneClinics* listing from the University of Washington, Seattle.
- Review of public and community health nurses survey results

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 challenges, which influence the efficiency and equity 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 have more than 75% of the state's populace and support the majority of the state's economic activity.

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 economy and low unemployment rate of approximately 2.6 percent (January, 2000), coupled with a highly coveted quality of life, has drawn a significant influx of new migrants to both urban and rural areas.

Colorado State Topography, with bisecting mountain range detail



Many 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 metropolitan area, which is located 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 groups. 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.

Population Demographics

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.

Colorado State Vital Statistics

The attached *Key Colorado Vital Statistics* (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 – 59,550 live births reported; birth rate of 14.7 per 1,000 population; general fertility rate of 63.7 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 6.7 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, cardiovascular 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/ps/mch/mchdatasets.html

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.

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 (CRCSN), Colorado's birth defects monitoring and prevention program. The purposes of CRCSN 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.

Colorado State Genetics Programs

Of particular value to the development of this plan is the ongoing coordination of activities, data, resource monitoring and education that 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

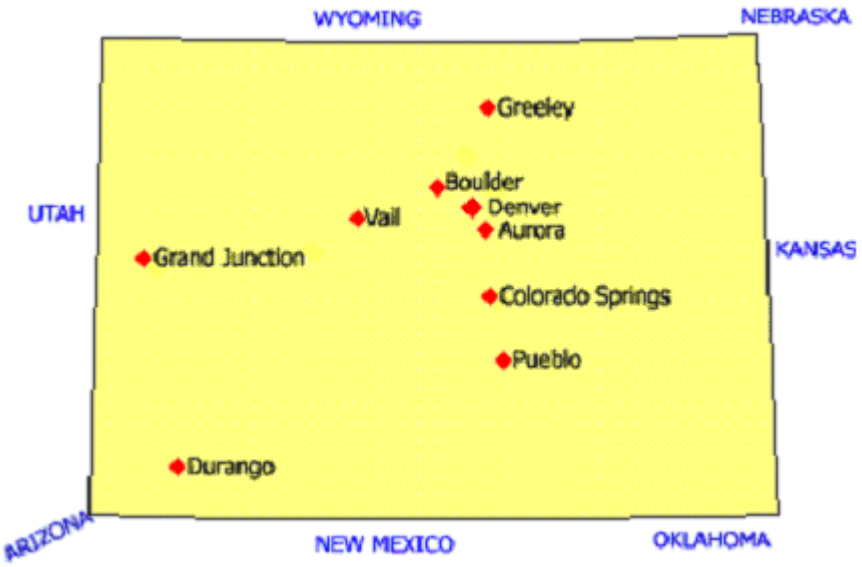
Direct Genetics Services

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 area. 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



Genetic counseling services are provided throughout the State of Colorado, primarily by the genetics unit 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.

Listing of Genetic Service Providers in Colorado

(See Appendix C: Genetics Services in the State of Colorado*)*

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 100,000 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). Dr. Bill Letson serves as medical director of the program. All newborns are screened by the heel stick blood spot method for PKU, biotinidase deficiency, galactosemia, hemoglobinopathies, hypothyroidism, congenital adrenal hyperplasia and cystic fibrosis. These screens are done at the CDPHE laboratory and the results downloaded to the Metabolic Screening Follow-up Program. 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 Appendix 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.

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.

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.

Indirect Genetics Services – Primary Care Providers and Nurses

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

- There are currently 1,802 Primary Care Physicians practicing in the State of Colorado
- There are 39 Public Health Nursing Agencies in Colorado 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.

Laboratory Services

Colorado benefits from the availability of excellent clinical laboratory services, which provide a spectrum of the diagnostic and prognostic test modalities currently available for various inherited and acquired diseases. The state maintains a public health laboratory 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, one biochemical genetics, and one 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

Current Utilization and Perceptions Among Key Medical Providers

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 Appendix F: Primary Care Providers Survey and

Appendix G: Public Nurses Survey, 2000.

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.

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.

Projected Need for Services by 2005

Priority Needs

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.

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 been identified (see Online Mendelian Inheritance In Man (http://www.nbci.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.

Gaps in Services

The challenges of providing equal access to genetics services emphasize the need to address gaps in service. In the provision services, education and policy, 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.

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.

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 sequela. 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.

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.

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.

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.

Access

The Colorado Health Information Dataset (CoHID) provides a queriable 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

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.

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

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 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.

Long Term Goals

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. 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.

Sort Term Goals

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.

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.

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

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.

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.

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.

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.

Action Items - Program Development and Coordination

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 by 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.

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.

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.

Action 3 – Medical Home

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.

Additionally, work on the development of the Medical Home has already been initiated by the formation of the Medical Home Advisory Group, consisting of physician representatives of Pediatrics, Family Practice, Medicaid, several payer organizations and Family Voices. The Health Care Program for Children with Special Needs (HCP) convened this group and is likewise an active participant and supporter of the project. Strong ties exist between HCP and Family Voices. MCH funds are used to help support the Colorado Chapter and input from Family Voices is solicited as appropriate. Consumer input is also available from consumer representatives of the Newborn Genetics Screening Advisory Committee and the existing State Genetics Advisory Committee. HCP has also recently hired a parent consultant who would be available to give input into the grant activities of the current project.

Plans have already been proposed in the Maternal Child Health Bureau Grant application (referred to immediately above) to initiate development of the medical home for Colorado's children with special health care needs population, beginning with children identified through the Newborn Metabolic Screening and Newborn Hearing Screening Programs**.** The State Genetics Plan would support the efforts of this program and propose expansion of the patient profiles with data generated through the extended metabolic screening program utilizing Tandem Mass Spectrometry described in (C).

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.

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).

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.

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.

Recommendations:

- Secure a full-time educator (preferably a board certified genetic counselor) for the Newborn Screening (hearing and metabolic testing) programs. This position would provide crossover education options for professionals and patients in various situations.
- 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.
- Develop relationships with insurers to provide education on reimbursement issues, i.e. out-of-state lab requirements, genetic counseling, etc.
- 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.
- 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.
- 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.
- Optimize links from the state website to other genetics-related resources on the web.

- Develop a brochure regarding the benefit and indications for clinical genetic services throughout the lifecycle to assist in educating insurers and policy makers.
- 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:

Practitioner Awareness
Primary Care Providers
Public Health Nurses
Private Health Nurses
Population Awareness
Preventive Services
Maternal and Child Health
Newborn Screening
Developmental Health
Adult Services
Health Promotion
Preconception Education
Adult Predisposition Awareness
Cross-Audience Education
Target Audience Segmentation
Adoption Workers

Affected Families
Allied Healthcare Professionals
Clergy
Clinical Genetics Professionals
General Public
Insurance Providers
Law and Policy Makers
Legal Professionals
Medical and Professional Association Leaders
News Media
Researchers and Institutional Review Boards
Support Groups
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.

APPENDIX A: WEB REFERENCES

Center for Disease Control, Office of Genetics and Disease Prevention. www.cdc.gov/genetics/

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

Online Mendelian Inheritance In Man www.nbci.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.

Mountain States Genetics Network www.mostgenes.org/.

GeneTests, a directory of clinical laboratories providing testing for genetic disorders. www.genetests.org

"Life After the Human Genome Map." *Genetic Engineering News*, Vol. 20, No. 14, August 2000.

"Will Genetics Revolutionize Medicine?" Holtzman NA and Marteau, TM *New England Journal of Medicine,* 2000 Jul 13; 343(2).

Enhancing the Oversight of Genetics Tests: Recommendations of the Secretary's Advisory Committee on Genetic Testing (SACGT). January 2000. National Institutes of Health. www4.od.nih.gov/oba/sacgt.htm

Colorado Department of Local Affairs. *Colorado State Population Projections, 1990-2025.* June 2000. www.dola.state.co.us/demog/project.htm

Comparing Colorado State Genetics Services to the CORN *Guidelines for Clinical Genetic Services for the Public's Health.* December 12, 2000. mchneighborhood.ichp.edu/pacnorgg.

APPENDIX B: BIBLIOGRAPHY

Angier, N., "Do Races Differ? Not Really, DNA Study Shows," New York Times, August 22, 2000.

Arizona Department of Health Services Office of Women's and Children's Health, "Genetic Health Care in Arizona: A Plan for the 21st Century," *Arizona Genetic Health Services Plan*, (Phoenix, Arizona, 1999).

Austin, Melissa A., Peyser, Patrica A., and Khoury Muin J., "The Interface of Genetics and Public Health: Research and Educational Challengers," Annual Review of Public Health.2000.21:81-99.

Barton, James C. and Ronald T. Acton, Inheritance of two *HFE* mutations in African Americans: Cases with hemochromatosis phenotypes and estimates of hemochromatosis phenotype frequency. *Genetics in Medicine*, 2001:3(4):294–300.

Brink, S. "Little-Used Newborn Screening Test Can Prevent Real Heartache," *US News and World Report*, website: *http://www.usnews.com/usnews/ issue/00017/nycu/babies.htm*, visited January 19, 2001.

Collins, Francis S., "Shattuck Lecture-Medical and Societal Consequences of the Human Genome Project," The New England Journal of Medicine, Vol.341:28-37 (July1,1999).

Colorado Department of Local Affairs, *Colorado State Population Projections, 1990-2005.* June 2000. http://www.dola.state.co.us/demog/project.htm, visited August 12-15, 2000; October 3, 12, 2000; November 4, 2000; March 15-17, 2001.

Colorado Department of Public Health & Environment. *Public and Community Health Nurses Survey*, 2000 (2000).

Cowley, Geoffrey, *Flunk the Test and Lose Your Insurance*, Newsweek, 48-50 (December 23, 1996).

D.A. Grimes and G.R. Snively, "Patient's Understanding of Medical Risks: Implications for Genetic Counseling," *Obstetrics and Gynecology* 93 (1999): 910-912.

Demography section, Colorado Division of Local Government. *Key Colorado Vital Statistics for 1998*. http://www.cdphe.state.co.us/hs/summaryweb98.pdf, visited August 12-15, 2000; October 3, 12, 2000.

Department of Health and Human Services. *Health Insurance in the Age of Genetics.* Report of the Secretary to the President. July 1997.

D. Wertz and R. Gregg, "Optimizing Genetics Services on a Social, Ethical, and Policy Context: Suggestions from Consumers and Providers in the New England regional Genetics Group," *The Genetic Resource 10*, no. 2 (1996): 49.

E.S. Lander, "Scientific Commentary: The Scientific Foundations and Medical and Social Prospects of the Human Genome Project," *Journal of Law, Medicine, and Ethics* 26 (1998): 184.

Barbara Fuller, J.D., National Action Plan on Brest Cancer, unpublished data.

Garland, Michael J., Experts and the public: a needed partnership for genetic policy," Public Understand. Sci.8(1999) 241-254.

Geller, Lisa N. et al, *Individual, Family, and Societal Dimensions of Genetic Discrimination : A Case Study Analysis*, Science and Engineering Ethics, Vol. 2, 71-88 (1996).

G. Geller and N.A. Holtzman, "A Qualitative Assessment of Primary Care Physician's Perceptions About the Ethical and Social Implications of Offering Genetics Testing," *Qualitative Health Research* 5 (1995): 1, 97.

Genetics Web Sites "Lists of Lists", Compiled by Stephen M. Modell.

C. Ghosh, "Medical Breakdown," *Forbes*, May 29, 2000, at Forbes website: ***http://www.forbes.com/forbes/00/0529/6513174a.htm***, visited January 24, 2001.

Green, Carol L., ed. "Integrating Genetics into State Chronic Disease Programs – Recommendations for Actions." A report from the Association of State and Territorial Chronic Disease Program Director's Genetics Retreat: December 2000.

Harris Poll, 1995 #34.

Susan J. Hayflick and M. Patrice Eiff, "Role of Primary Care Providers in the Delivery of Genetics Services," *Community Genetics*, Departments of Molecular and Medical Genetics, Pediatrics, Family Medicine, Oregon Health Sciences University, Portland, Oregon (1998): 18-22.

Susan J. Hayflick and M. Patrice Eiff, "Role of Primary Care Providers in the Delivery of Genetics Services," *Community Genetics*, Departments of Molecular and Medical Genetics, Pediatrics, Family Medicine, Oregon Health Sciences University, Portland, Oregon (1998): 18-22.

Hepburn, "Genetic Testing and Early Diagnosis," 109; A. Lippman, "Prenatal Genetic Testing and Screening: Constructing Needs and Reinforcing Inequities," *American Journal of Law and Medicine* 17 (1991): 15, 25.

N.A. Holtzman and L.B. Andrews, "Ethical and Legal Issues in Genetic Epidemiology," *Epidemiological Reviews* 19 (1997): 163, 169.

N.A. Holtzman, "Medical and Ethical Issues in Genetic Screening – An Academic View," *Environmental Health Perspectives* 104 (1996): 987.

Holtzman, Neil A., Watson, Michael S., "Promoting Safe and Effective Genetic Testing in the United States: Final Report of the Task Force on Genetic Testing," Executive Summary, The National Human Genome Research Institute, September 1997.

Institute of Medicine Committee on Assessing Genetic Risks, *Assessing Genetic Risks: Implications for Health and Social Policy*, ed. L.B. Andrews et al. (Washington, D.C.: National Academy Press, 1994): 4, 65.

Khoury, Muin J., Wylie, Burke, Thompson, Elizabeth, "Genetics and Public Health: A Framework for Integration of Human Genetics into Public Health Practice",http://www.cdc.gov/genetics/publications/21stcentury_1.htm, August 11,2000.

Khoury, Muin,: "Genetic Susceptibility to Birth Defects in Humans: From Gene Discovery to Public Health Action", http://www.cdc.gov/genetics/publications/bthdefect.htm, August 11,2000.

Khoury,MuinJ., Thrasher, James, F., Burke, Wylie, Gettig, Elizabeth A., Fridinger, Fred and Jackson, Richard, "Challenges in Communicating Genetics: A Public Health Approach," Genetics in Medicine, May/June 2000 Vol.2 No. 3, http://www.cdc.gov/genetics/publications/communicate.htm

Kolata, Gina, *Advent of Testing for Breast Cancer Genes Leads to Fears of Disclosure and Discrimination*, New York Times, C1 (February 4, 1997).

Lapman, E. Virginia et al, *Genetic Discrimination : Perspectives of Consumers*, Science, Vol. 274, 621-24 (October 25, 1996).

H. Markel, "Scientific Advances and Social Risks: Historical Perspectives of Genetic Screening Programs for Sickle Cell Disease, Tay-Sachs Disease, Neural Tube Defects and Down Syndrome: in NIL-DOE Task Force on Genetic Testing, *Promoting Safe and Effective Genetic Testing*, 161, 163-164; Chapter 5, page 115; Chapter 10, page 281-283.

Miller, Paul S., *Is There a Pink Slip in My Genes? Genetic Discrimination in the Workplace*, 3 University of Maryland, Journal of Health Care and Law and Policy 224 (to be published Summer,2000).

Mittman, I.S. and Secundy, M.G. (1998a). Genetic Education to Diverse Communities Employing a Community Empowerment Model. Community Genetics,1(3),160-165.

Mittman, I.S. and Secundy, M.G. (1998b). A National Dialogue on Genetics and Minority Issues. Community Genetics, 1(3),190-200.

T.H. Murray, "Genetic Exceptionalism and Future Diaries: Is Genetic Information Different from Other Medical Information?" in *Genetic Secrets: Protecting Privacy and Confidentiality in a Genetic Era*, ed. M. A. Rothstein (New Haven, CT: Yale University Press, 1997): 60.

Michigan Commission on Genetic Privacy and Progress: Final Report and Recommendations, "Summary of Genetic Commission Recommendations," February 1999.

National Institutes of Health-U.S. Department of Energy Working Group on Ethical, Legal, and Social Implications of Human Genome Research Task Force on Genetic Information and Insurance, *Genetic Information and Health Insurance* (Washington, D.C.: National Institutes of Health, 1993) National Human Genome Research Institute website: ***http://www.nghri.gov/About_NHGRI/Der/Elsi/itf.html***, visited December 12, 2000.

National Institutes of Health-Secretary's Advisory Committee on Genetic Testing, *A Public Consultation on Oversight of Genetic Tests* (Bethesda, Maryland: National Institutes of Health, 2000): 9,18, 21, 24, 29-33.

National Institutes of Health-Department of Energy (NIH-DOE) Working Group on Ethical, Legal and Social Implications of Human Genome Research Task Force on Genetic Testing, *Promoting Safe and Effective Genetic Testing*, ed. N.A. Holtzman and M.S. Watson (Washington, D.C.: National Institutes of Health, 1997), 42-62.

New York State Task Force on Life and the Law, *Genetic Testing and Screening in the Age of Genomic Medicine*, Carl H. Coleman, J.D., exec. dir. (New York, NY: New York State Task Force on Life and the Law, 2000), 3, 11,60,-62, 79,91, 104165, 187,389, 392.

N. Press and C.H. Browner, "Risk, Autonomy, and Responsibility: Informed Consent for Prenatal Testing," *Hastings Center Report* 25, Special Supplement (1995): S9, S10.

National Institutes of Health *Enhancing the Oversight of Genetics Tests: Recommendations of the Secretary's Advisory Committee on Genetic Testing (SACGT)*. (Washington, D.C., January, 2000). National Institutes of Health website: http:// www4.od.nih.gov/oba/sacgt.htm visited August 5, 2000, December 13, 2000.

Pernick, Martin, S.," Eugenics and Public Health in American History," American Journal of Public Health 87 (November 1997), 1767-1772.

Paul G. Ramsey, MD, J. Randall Curtis, MD, MPH, Douglas S. Paauw, MD, Jan D. Carline, PhD, Marjorie D. Wenrich, MPH, "History-taking and Preventive Medicine Skills among Primary Care Physicians: An Assessment Using Standardized Patients," ***The American Journal of Medicine,*** 104:2 : 152-158.

Rothenberg, Karen H., *Genetics Information and Health Insurance: State Health Approaches,* Journal of Law, Medicine and Ethics, Vol. 23:4, 312-19 (Winter 1995).

Stopfer, J. "To Test or Not to Test? Genetic Counseling is the Key." University of Pennsylvania Cancer Risk Evaluation Programs Posted Date: January 11, 1998.

N. Touchette, N. A. Holtzman, J. G. Davis, S. Feetham, *Toward the 21st Century : Incorporating Genetics into Primary Health Care.* (New York, NY: Cold Spring Harbor Laboratory Press). 1997.

Washington State Department of Health, *Washington State Genetics Education Plan*, Evans Group, (Seattle, Washington, 1997).

Weijer C. and E.J Emanuel (2000) Protecting Communities in Biomedical Research. Science 289: 1142-1144.

APPENDIX C: GENETICS SERVICES IN THE STATE OF COLORADO

Colorado Springs – Memorial Hospital Pediatrics Specialties Clinic
2121 East La Salle
Colorado Springs, CO 80909
(719) 365-2244
On site scheduling, clinics held monthly

Denver – The Children's Hospital
Clinical Genetics, B-300
1056 E. 19th Avenue
Denver, CO 80218
(303) 861-6322
Clinics held regularly

Denver – The University of Colorado Health Sciences Center
Adult Medical Genetics Program
12635 East Montview Boulevard, Suite 125
Aurora, CO 80010
(303) 724-0797

Durango – San Juan Basin Health Department
281 Sawyer Drive
Durango, CO
(970) 247-5702
Scheduling through Clinical Genetics and Metabolism department at The Children's Hospital, (303) 861-6395
Two clinics held annually – one in spring, one in fall

Grand Junction – Mesa County Health Department
510 29 ½ Road
Grand Junction, CO 81504
(970) 248-6906
Scheduling through Clinical Genetics and Metabolism department at The Children's Hospital, (303) 861-6395
Clinics held February, April, June, August, October, December

Greeley – Weld County Health Department
1555 North 17th Avenue
Greeley, CO
(970) 304-6420
Scheduling through Clinical Genetics and Metabolism department at The Children's Hospital, (303) 861-6395
Clinics held January, March, May, July, September and November

Pueblo – Pueblo County Health Department
151 Center Main
Pueblo, CO
(719) 583-4369
On site scheduling – clinics held February, April, June, August, October, December

APPENDIX D: STATE POPULATION PROJECTIONS, 1990-2002

Colorado Department of Local Affairs

Table 3. FINAL COLORADO POPULATION ESTIMATES BY COUNTY, 1990 - 2002

COUNTIES	USCB Count April, 1990	USCB Count April, 2000	CDS Est. July, 2000	CDS Est. July, 2001	CDS Est. July, 2002	Average Annual Percent Change 1990 - 00	2000 - 01	2001 - 02
COLORADO	3,294,473	4,301,997	4,335,540	4,441,377	4,516,847	2.7%	2.4%	1.7%
Adams	258,316	347,961	350,642	361,262	375,380	3.0%	3.0%	3.9%
Alamosa	13,617	14,966	15,139	15,282	15,377	0.9%	0.9%	0.6%
Arapahoe	391,511	488,896	491,143	503,846	513,965	2.2%	2.6%	2.0%
Archuleta	5,345	9,898	10,028	10,548	10,912	6.4%	5.2%	3.5%
Baca	4,556	4,517	4,516	4,514	4,401	-0.1%	-0.1%	-2.5%
Bent	5,048	5,998	5,971	5,865	6,072	1.7%	-1.8%	3.5%
Boulder	208,949	269,785	271,051	275,809	277,601	2.6%	1.8%	0.6%
Broomfield	24,638	39,199	39,466	40,621	41,948	4.8%	2.9%	3.3%
Chaffee	12,684	16,242	16,298	16,522	16,692	2.5%	1.4%	1.0%
Cheyenne	2,397	2,231	2,230	2,228	2,207	-0.7%	-0.1%	-0.9%
Clear Creek	7,619	9,322	9,391	9,485	9,528	2.0%	1.0%	0.5%
Conejos	7,453	8,400	8,400	8,401	8,400	1.2%	0.0%	0.0%
Costilla	3,190	3,663	3,675	3,723	3,746	1.4%	1.3%	0.6%
Crowley	3,946	5,518	5,513	5,491	5,822	3.4%	-0.4%	6.0%
Custer	1,926	3,503	3,540	3,686	3,769	6.2%	4.1%	2.3%
Delta	20,980	27,834	28,009	28,709	29,196	2.9%	2.5%	1.7%
Denver	467,610	553,693	555,782	560,365	560,882	1.7%	0.8%	0.1%
Dolores	1,504	1,844	1,844	1,844	1,876	2.1%	0.0%	1.7%
Douglas	60,391	175,766	180,690	200,385	213,526	11.3%	10.9%	6.6%
Eagle	21,928	41,659	43,354	44,824	45,819	6.6%	3.4%	2.2%
Elbert	9,646	19,872	20,188	21,453	21,936	7.5%	6.3%	2.3%
El Paso	397,014	516,929	520,572	533,526	541,069	2.7%	2.5%	1.4%
Fremont	32,273	46,145	46,439	47,209	47,431	3.6%	1.7%	0.5%
Garfield	29,974	43,791	44,267	46,173	47,441	3.9%	4.3%	2.7%
Gilpin	3,070	4,757	4,775	4,845	4,899	4.5%	1.5%	1.1%
Grand	7,966	12,442	12,884	13,253	13,421	4.6%	2.9%	1.3%
Gunnison	10,273	13,956	13,967	14,012	13,999	3.1%	0.3%	-0.1%
Hinsdale	467	790	791	794	810	5.4%	0.4%	2.0%
Huerfano	6,009	7,862	7,861	7,857	8,034	2.7%	-0.1%	2.3%
Jackson	1,605	1,577	1,586	1,620	1,603	-0.2%	2.2%	-1.0%
Jefferson	436,908	525,330	526,269	529,404	530,821	1.9%	0.6%	0.3%
Kiowa	1,688	1,622	1,617	1,598	1,574	-0.4%	-1.2%	-1.5%
Kit Carson	7,140	8,011	8,012	8,007	8,034	1.2%	-0.1%	0.3%
Lake	6,007	7,812	7,908	7,878	7,902	2.7%	-0.4%	0.3%
La Plata	32,284	43,941	44,566	45,616	46,281	3.1%	2.4%	1.5%

Table 3. FINAL COLORADO POPULATION ESTIMATES BY COUNTY, 1990 - 2002

COUNTIES	USCB Count April, 1990	USCB Count April, 2000	CDS Est. July, 2000	CDS Est. July, 2001	CDS Est. July, 2002	Average Annual Percent Change 1990 - 00	2000 - 01	2001 - 02
Larimer	186,136	251,494	253,137	259,707	262,711	3.1%	2.6%	1.2%
Las Animas	13,765	15,207	15,276	15,550	15,836	1.0%	1.8%	1.8%
Lincoln	4,529	6,087	6,170	6,117	6,123	3.0%	-0.9%	0.1%
Logan	17,567	20,574	20,862	21,920	21,917	1.6%	5.1%	0.0%
Mesa	93,145	116,935	117,656	119,961	122,463	2.3%	2.0%	2.1%
Mineral	558	831	833	843	865	4.1%	1.2%	2.6%
Moffat	11,357	13,184	13,185	13,190	13,288	1.5%	0.0%	0.7%
Montezuma	18,672	23,830	23,864	23,999	24,216	2.5%	0.6%	0.9%
Montrose	24,423	33,432	33,666	34,601	35,435	3.2%	2.8%	2.4%
Morgan	21,939	27,171	27,261	27,623	27,854	2.2%	1.3%	0.8%
Otero	20,185	20,311	20,244	19,976	19,717	0.1%	-1.3%	-1.3%
Ouray	2,295	3,742	3,771	3,888	3,991	5.0%	3.1%	2.6%
Park	7,174	14,523	14,703	15,325	15,738	7.3%	4.2%	2.7%
Phillips	4,189	4,480	4,486	4,511	4,529	0.7%	0.6%	0.4%
Pitkin	12,661	14,872	15,913	16,197	16,257	1.6%	1.8%	0.4%
Prowers	13,347	14,483	14,434	14,240	14,180	0.8%	-1.3%	-0.4%
Pueblo	123,051	141,472	142,054	144,383	147,057	1.4%	1.6%	1.9%
Rio Blanco	6,051	5,986	5,986	5,986	6,063	-0.1%	0.0%	1.3%
Rio Grande	10,770	12,413	12,434	12,518	12,559	1.4%	0.7%	0.3%
Routt	14,088	19,690	20,102	20,551	20,941	3.4%	2.2%	1.9%
Saguache	4,619	5,917	5,954	6,100	6,195	2.5%	2.5%	1.6%
San Juan	745	558	558	560	563	-2.8%	0.3%	0.5%
San Miguel	3,653	6,594	6,666	6,956	7,135	6.1%	4.3%	2.6%
Sedgwick	2,690	2,747	2,742	2,722	2,743	0.2%	-0.7%	0.8%
Summit	12,881	23,548	25,725	26,355	26,798	6.2%	2.4%	1.7%
Teller	12,468	20,555	21,145	21,827	21,988	5.1%	3.2%	0.7%
Washington	4,812	4,926	4,920	4,898	5,071	0.2%	-0.5%	3.5%
Weld	131,817	180,862	183,557	194,318	202,329	3.2%	5.9%	4.1%
Yuma	8,954	9,841	9,853	9,900	9,911	0.9%	0.5%	0.1%

APPENDIX E: KEY COLORADO VITAL STATISTICS FOR 1998

Key Colorado Vital Statistics for 1998

- **Live Births — 59,550; Birth Rate — 14.7 per 1,000 population; General Fertility Rate — 63.7 per 1,000 women age 15-44**

  The number of live births was up from 56,505 in 1997.

- **Teen Births (age 10-14) — 122; Teen Births (age 15-19) — 7,057**

  The fertility rate for the youngest mothers (age 10-14) increased slightly from 0.7 in 1997 to 0.8 in 1998. The fertility rate for teens age 15-19 increased from 47.1 in 1997 to 48.4 in 1998.

- **Low Weight Births — 5,168 or 8.7 percent**

  The percent of low weight births (<2,500 grams) decreased from 8.9 in 1997 to 8.7 in 1998. Colorado has maintained a higher percent of low weight births than the nation since at least 1950. The U.S. reported 7.6 percent low weight births in 1998.

- **Births to Unmarried Women — 15,181 or 25.5 percent**

  The percentage of births to unmarried mothers rose from 25.2 percent in 1997 to 25.5 percent in 1998. The U.S. continued to report a higher percent of births to unmarried mothers than Colorado at 32.8 percent in 1998.

- **Marriages — 33,824; Marriage Rate — 8.3 per 1,000 population**

  The number of marriage applications received in the state decreased from 34,883 in 1997,while the rate of applications per 1,000 population decreased from 8.8 in 1997.

- **Marriages Dissolutions — 19,537; Marriage Dissolution Rate — 4.8 per 1,000 population**

- **Deaths — 26,582; Crude Death Rate — 655.6 per 100,000 population; Age-Adjusted Death Rate — 426.7 per 100,000 population**

  The number of resident deaths increased from 25,606 in 1997, and the crude death rate increased from 650.7 per 100,000 population in 1997. The age-adjusted death rate increased from 424.0 per 100,000 population in 1997. Colorado has consistently reported lower death rates than the nation overall. The U.S. reported a crude rate of 865.0 per 100,000 population and an age-adjusted rate of 470.7 per 100,000 population in 1998.

- **Crude Death Rates for Ten Leading Causes of Death in Colorado**

  The ten leading causes of death and their associated crude death rates per 100,000 population were heart disease— 163.0, cancer 143.2, COPD 45.2, cerebrovascular disease 42.8, unintentional injuries 37.2, pneumonia/influenza 25.5, suicide 14.8, diabetes 14.0, atherosclerosis 10.2, and Alzheimer's disease 9.3. Among the five leading causes of death, Colorado reported lower crude death rates than the U.S. for heart disease, cancer, and cerebrovascular disease, and higher crude death rates for COPD and unintentional injuries. Colorado also reported higher crude rates than the U.S. for suicide and atherosclerosis.

- **Infant Deaths — 396, Infant Mortality Rate — 6.7 per 1,000 live births**

  Following a decreasing trend over the past 55 years, the state's infant mortality rate was slightly higher at 6.7than the record low of 6.5 in 1995. In preliminary data, the U.S. reported a record low rate of 7.2 in 1998.

- **Spontaneous Fetal Deaths of 20+ Weeks Gestation — 376**

  Reports of spontaneous fetal deaths of at least 20 weeks gestation decreased from 389 in 1997.

- **Induced Terminations of Pregnancy — 7,493**

  The number of induced terminations of pregnancy reported as occurring in Colorado (residents and nonresidents) decreased from 9,183 in 1997. A total of 6,657 induced terminations occurred among Colorado residents in 1998.

- **Population — 4,054,343**

  Colorado's population continued to grow with an increase of 2.5 percent from 1997.

NOTE: Numbers, rates, and percentages are based on events occurring to Colorado residents only except for marriage data, which are based on all occurrences within Colorado (residents and nonresidents) unless otherwise noted. Population data are provided by the Demography Section, Colorado Division of Local Government (formerly the Demographic Section, Colorado Department of Local Affairs).

APPENDIX F: PRIMARY CARE PROVIDERS SURVEY

APPENDIX G: PUBLIC NURSES SURVEY, 2000

APPENDIX H: CURRENT LISTING OF HEREDITARY DISEASES

21-Hydroxylase Deficiency
22q11.2 Deletion Syndrome

3-M Syndrome

ARSACS
ATP7A-Related Copper Transport Disorders
Aceruloplasminemia
Achondrogenesis Type 1B
Achondroplasia
Adrenal Hypoplasia Congenita, X-Linked
Adrenoleukodystrophy, X-Linked
Agammaglobulinemia, X-Linked
Alagille Syndrome
Alexander Disease
Alkaptonuria
Alpha-Mannosidosis
Alpha-Thalassemia X-Linked Mental Retardation Syndrome
Alport Syndrome
Alstrom Syndrome
Alzheimer Disease Overview
Alzheimer Disease, Early-Onset Familial
Amish Lethal Microcephaly
Amyotrophic Lateral Sclerosis Overview
Androgen Insensitivity Syndrome
Angelman Syndrome
Aniridia
Anophthalmia / Microphthalmia Overview
Ataxia Overview
Ataxia with Oculomotor Apraxia Type 1
Ataxia-Telangiectasia
Atelosteogenesis Type 2
Autism Overview

BRCA1 and BRCA2 Hereditary Breast/Ovarian Cancer
Bardet-Biedl Syndrome
Basal Ganglia Calcification, Familial Idiopathic
Beckwith-Wiedemann Syndrome
Berardinelli-Seip Congenital Lipodystrophy
Best Vitelliform Macular Dystrophy
Beta-Thalassemia
Biotinidase Deficiency
Branchiootorenal Syndrome
Breast Cancer Genetics Overview

CADASIL
Canavan Disease
Carney Complex
Cerebral Cavernous Malformation, Familial
Cerebrotendinous Xanthomatosis
Char Syndrome
Charcot-Marie-Tooth Overview
Charcot-Marie-Tooth Type 1
Charcot-Marie-Tooth Type 2
Charcot-Marie-Tooth Type 2E/1F
Charcot-Marie-Tooth Type 4
Charcot-Marie-Tooth Type 4A
Charcot-Marie-Tooth Type X
Childhood Ataxia with Central Nervous System Hypomelination/Vanishing White Matter
Choreoacanthocytosis
Choroideremia
Cockayne Syndrome
Coffin-Lowry Syndrome
Congenital Central Hypoventilation Syndrome
Congenital Contractural Arachnodactyly
Congenital Fibrosis of the Extraocular Muscles
Congenital Ichthyosis, Autosomal Recessive
Congenital Muscular Dystrophy Overview
Congenital Myasthenic Syndromes
Craniosynostosis Syndromes, FGFR-Related
Cystic Fibrosis
Cystinosis

DRPLA
Deafness and Hereditary Hearing Loss Overview
Deafness-Dystonia-Optic Neuronopathy Syndrome
Dopa-Responsive Dystonia
Dopamine Beta-Hydroxylase Deficiency
Dysferlinopathy
Dystonia Overview
Dystonia, Early-Onset Primary (DYT1)
Dystrophinopathies

ELA2-Related Neutropenia
Ehlers-Danlos Syndrome, Classic Type
Ehlers-Danlos Syndrome, Kyphoscoliotic Form
Ehlers-Danlos Syndrome, Vascular Type
Enlarged Parietal Foramina/Cranium Bifidum
Epidermolysis Bullosa Simplex
Episodic Ataxia Type 2

Fabry Disease
Facioscapulohumeral Muscular Dystrophy
Factor V Leiden Thrombophilia
Familial Adenomatous Polyposis
Familial Dysautonomia
Familial Hemiplegic Migraine
Familial Hyperinsulinism (FHI)
Familial Mediterranean Fever
Fanconi Anemia
Fragile X Syndrome
Free Sialic Acid Storage Disorders
Friedreich Ataxia
Frontotemporal Dementia with Parkinsonism-17

Galactosemia
Gaucher Disease
Giant Axonal Neuropathy
Glucose Transporter Type 1 Deficiency Syndrome
Glycine Encephalopathy
Greig Cephalopolysyndactyly Syndrome

HFE-Associated Hereditary Hemochromatosis
Hemophilia A
Hemophilia B
Hereditary Diffuse Gastric Cancer
Hereditary Hemorrhagic Telangiectasia
Hereditary Neuropathy with Liability to Pressure Palsies
Hereditary Non-Polyposis Colon Cancer
Hereditary Sensory Neuropathy Type I
Hereditary Spastic Paraplegia Overview
Hermansky-Pudlak Syndrome
Hexosaminidase A Deficiency
Hirschsprung Disease Overview
Holoprosencephaly Overview
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency
Huntington Disease
Huntington Disease-Like 2
Hutchinson-Gilford Progeria Syndrome
Hyperkalemic Periodic Paralysis Type 1
Hyperoxaluria, Primary, Type 1
Hypochondroplasia
Hypohidrotic Ectodermal Dysplasia

Hypokalemic Periodic Paralysis

IRF6-Related Disorders
Inclusion Body Myopathy 2
Incontinentia Pigmenti

Jervell and Lange-Nielsen Syndrome
Joubert Syndrome
Juvenile Polyposis Syndrome

Krabbe Disease

L1 Syndrome
Leber Hereditary Optic Neuropathy
Lenz Microphthalmia Syndrome
Lesch-Nyhan Syndrome
Li-Fraumeni Syndrome
Limb-Girdle Muscular Dystrophy Overview
Lipoprotein Lipase Deficiency, Familial
Lowe Syndrome
Lymphoproliferative Disease, X-Linked

MELAS
MERRF
Malignant Hyperthermia Susceptibility
Marfan Syndrome
McKusick-Kaufman Syndrome
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
Megalencephalic Leukoencephalopathy with Subcortical Cysts
Mitochondrial DNA Deletion Syndromes
Mitochondrial DNA-Associated Leigh Syndrome and NARP
Mitochondrial Disorders Overview
Mucopolysaccharidosis Type I
Multiminicore Disease
Multiple Endocrine Neoplasia Type 2
Multiple Epiphyseal Dysplasia, Autosomal Dominant
Multiple Epiphyseal Dysplasia, Recessive
Multiple Exostoses, Hereditary
Myoclonus-Dystonia
Myotonic Dystrophy
Myotubular Myopathy, X-Linked

NDP-Related Retinopathies
Nail-Patella Syndrome
Nemaline Myopathy
Nephrogenic Diabetes Insipidus
Neurofibromatosis 1
Neurofibromatosis 2
Neuronal Ceroid-Lipofuscinosis
Nevoid Basal Cell Carcinoma Syndrome
Niemann-Pick Disease, Type C
Nijmegen Breakage Syndrome
Nocturnal Frontal Lobe Epilepsy, Autosomal Dominant
Nonsyndromic Hearing Loss and Deafness, DFNA3
Nonsyndromic Hearing Loss and Deafness, DFNB1
Noonan Syndrome

Ocular Albinism, X-Linked
Oculocutaneous Albinism Type 1
Oculocutaneous Albinism Type 2
Oculopharyngeal Muscular Dystrophy
Oral-Facial-Digital Syndrome Type I
Organic Acidemias Overview

PLOSL
PLP1-Related Disorders
PROP1 - Related Combined Pituitary Hormone Deficiency
PTEN Hamartoma Tumor Syndrome
Pallister-Hall Syndrome
Pantothenate Kinase-Associated Neurodegeneration
Parkin Type of Juvenile Parkinson Disease
Parkinson Disease Overview
Pendred Syndrome
Periventricular Heterotopia, X-Linked
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum
Peutz-Jeghers Syndrome
Phenylalanine Hydroxylase Deficiency
Polycystic Kidney Disease, Autosomal Dominant
Polycystic Kidney Disease, Autosomal Recessive
Prader-Willi Syndrome
Primary Pulmonary Hypertension
Prion Diseases
Progressive Familial Intrahepatic Cholestasis
Pseudoxanthoma Elasticum
Pyridoxine-Dependent Seizures

Retinitis Pigmentosa Overview
Retinoblastoma
Rett Syndrome
Rhizomelic Chondrodysplasia Punctata Type 1
Romano-Ward Syndrome
Rothmund-Thomson Syndrome
Rubinstein-Taybi Syndrome
Russell-Silver Syndrome

SOST-Related Sclerosing Bone Dysplasias
SPG4
Saethre-Chotzen Syndrome
Schimke Immunoosseous Dysplasia
Sialuria
Sickle Cell Disease
Smith-Lemli-Opitz Syndrome
Smith-Magenis Syndrome
Spinal Muscular Atrophy
Spinal and Bulbar Muscular Atrophy
Spinocerebellar Ataxia Type 1
Spinocerebellar Ataxia Type 2
Spinocerebellar Ataxia Type 3
Spinocerebellar Ataxia Type 6
Spinocerebellar Ataxia Type 7
Spinocerebellar Ataxia Type 8
Spinocerebellar Ataxia Type10
Spondyloepiphyseal Dysplasia Tarda, X-Linked
Stickler Syndrome
Succinic Semialdehyde Dehydrogenase Deficiency

Thanatophoric Dysplasia
Thiamine-Responsive Megaloblastic Anemia Syndrome
Thoracic Aortic Aneurysms and Aortic Dissections
Transthyretin Amyloidosis
Tuberous Sclerosis Complex

Urea Cycle Disorders Overview
Usher Syndrome Type I
Usher Syndrome Type II

Von Hippel-Lindau Syndrome

Waardenburg Syndrome Type 1
Werner Syndrome
Williams Syndrome
Wilms Tumor Overview
Wilson Disease
Wolf-Hirschhorn Syndrome

X-Linked Juvenile Retinoschisis
X-Linked Severe Combined Immunodeficiency
XX Male Syndrome
Xeroderma Pigmentosum

Y Chromosome Infertility

APPENDIX I: LETTER FROM STATE PUBLIC HEALTH OFFICIAL

re: ability to sustain efforts stimulated by these funds after the project period ends

APPENDIX J: LETTERS OF SUPPORT

APPENDIX K: BIOGRAPHICAL SKETCHES

Name (Last, First, MI) Letson, George, William	Title MCH Medical Epidemiologist	Birth Date (Month/Day/Year) 10-10-51

Education
(Begin with Baccalaureate or other initial professional education and include postdoctoral training)

Institution and Location	Degree	Year Completed	Field of Study
University of Colorado Biology	BA	1973	Environmental
Colorado State University	MS	1978	Zoology
University of Colorado School of Medicine	MD	1981	

Honors
Chief Resident in Pediatrics, University of Arizona School of Medicine
Fellow in Pediatrics Infectious Diseases, Johns Hopkins University

Research and Professional Experience
(List in reverse chronological order previous employment and experience. List in reverse chronological order most representative publications)

1997-present Maternal Child Health Consultant to Colorado Department of Public Health and Environment.
Wyoming State Health Officer and Administrator of the Division of Public Health, Wyoming Health Department.
Medical Epidemiologist with Centers for Disease Control and Prevention, Division of Viral and Rickettsial Diseases, Hepatitis Branch.
Medical Epidemiologist Centers for Disease Control and Prevention, Division of Vector –Borne Viral Diseases, Fort Collins, Colorado.
Director, Alaska Hib vaccine efficacy trial at Centers for Disease Control and Prevention, Arctic Investigations Program, Anchorage, Alaska

Letson GW, Hammerton S, Miller D. Lembitz D. Teen pregnancy reduction in program with peer staff. *J Adol Health* 2000; 26:240-1
Letson GW, Little JR, Ottman J, Miller GL Meningococcal vaccine in pregnancy: An assessment of infant risk. *Pediatr Infect Dis J* 1998; 17:26 1-3.
Wera T. Cochrane J, Hadden JC, Letson GW. The Larimer County Children's Clinic: A public/private partnership to provide medical care assess to indigent children. *Arch Pediatr Adolesc Med* 1994; 148:572-7.
Letson GW, Tsai TF, Bailey R. Eastern Equine Encephalitis (EEE). A description of the 1989 outbreak, recent epidemiologic trends and the association of rainfall with EEE occurrence. *Am J Trop Med Hyg* 1993; 49:677-85.

Tsai TF, Paul RM, Lynberg MC, :Letson GW. Congenital Yellow Fever viral infection after immunization in pregnancy. *J Infec Dis* 1993; 168:1520-3.
Letson GW, Gellin BG, Bulkow LR, Parks DJ, Ward JI. Severity and frequency of sequelae following meningitis in Alaska Native infants: Correlation with a scoring system for severity of sequelae. *Am J Dis Child* 1992; 146:560-6.
Bulkow LR, Wainwright RB, Letson GW, Chang SJ, Ward JI. Immunogenicity of four Haemophilus influnzae type b vaccines in Alaska Native infants. *Pediatr Infec Dis J* 1993; 12:484-92.
Santosham M, Letson GW, Wolff M, Reid R, Gahagan S, Adams R, Callahan C, Sack RB, Kapikian AZ. A field study of the safety and efficacy of two candidates. Rotavirus vaccines in a Native American population. *J Infec Dis* 1991; 163:483-7
Davidson M, Letson GW, Ward JI, Ball A, Bulkow L, Christenson P, Cherry JD. DTP immunization and susceptibility to infections: Is there a relationship? *Am J Dis Child* 1991; 145:750-4.
Ward JI, Brenneman G, Letson GW, Heyward W, and Alaska Vaccine Study Group. Limited protective efficacy of an Haemophilus influenzae type b conjugate vaccine (PRP-D) in young infants: Results of a trial in Alaska Native infants immunized at 2, 4, and 6 months of age. *N Engl J Med* 1990; 323:1393-1401.
Barton LL, Dawson JE, Letson GW, Luisiri A, Scalzo AJ. Simultaneous Erlichiosis and Lyme Disease. *Pediatr Infect Dis* J 1990; 9:127-9.
Letson GW, Santosham M, Reid R, Priehs C, Burns B, Jahnke A, Gahagan S, Nienstadt L, Johnson C, Smith D, Siber G. Comparison of active and combined passive/active immunization of Navajo children against *Haemophilus influenzae type b. Pediatr Infect Dis J* 1988; 7:747-52.
Santosham M, Burns, B, Reid R, Letson GW, Duncan B, Powlesland J, Foster S,. Garrett S, Croll L, Marshall W, Almeido-Hill J, Sack RB. Glycine-based oral rehydration solution: Reassessment of safety and efficacy. *J Pediatr* 1986; 109:795-801.

KATHLEEN D. WATTERS

2461 S. Ivanhoe Pl.
Denver, Colorado 80222
(303)753-9175

Business Address: Health Care Program for Children with Special Needs
Prevention and Intervention Services for Children and Youth
Colorado Department of Public Health and Environment
4300 Cherry Creek Drive South
Denver, Colorado 80222-1530
(303) 692-2418

Education:

1999 Regional Institute for Health and Environmental Leadership
University of Denver
Denver, Colorado

1980 MA, Audiology and Communication Disorders
University of Colorado
Boulder, Colorado

1977 BA, Speech/Language Pathology
University of Cincinnati
Cincinnati, Ohio

Professional Experience:

6/00-Present Acting Director, Health Care Program for Children with Special Needs (HCP)

11/99-6/00 Assistant Director, Health Care Program for Children with Special Needs (HCP)

3/91-11/99 Director, Community Consultation Team, Health Care Program for Children with Special Needs, Colorado Department of Public Health and Environment, Denver, Colorado (General Professional VI)

Manage multiple sub-programs and budgets within the HCP program through subordinate staff. Manage and supervise a team of 9 state specialty consultants (including Audiology, Deaf Education, Family Advocacy, Nursing, Nutrition, OT/PT, Speech, Social Work) who are leaders in their fields and have statewide responsibilities for building community-based service systems as well as managing program contracts with HCP regional offices. Develop, negotiate grant funding for special projects that enhance systems building and program goals. Responsible for legislative process activities that relate to the above specialty fields. Provide statewide training, public speaking, and interagency group process facilitation. Provide national leadership to the MCH Speech and Hearing organization. Responsible for program community development and interagency collaboration. Acts as an assistant/advisor to the program director in the areas of budget planning, program planning, needs

assessment, and systems building. Represents the Division MCH Programs on a statewide advisory for Traumatic Brain Injury and Co-chairs the subcommittee for children and adolescents.

6/88-3/91 Director, Hearing and Speech Services, Handicapped Children's Program, Colorado Department of Public Health (Program Administrator I)

Directly supervised 4 professional and 1 support staff. Managed a budget of approximately .75 million. Responsibilities included program planning, development of statewide policy and standards of care in the areas of hearing and speech, research, data management, staff development, grant management and program evaluation.

10/82-6/88 Director, Parent-Home Training Program, Handicapped Children's Program, Colorado Department of Public Health, Denver, Colorado (Audiologist IV)

Managed a statewide early intervention program including budget management**,** training workshops, small consultative staff and advisory, small grants and contracts.

10/80-10/82 Part-time Private Practice, Audiology/Aural Habilitation, Colorado Springs and Denver, Colorado

12/79-10/82 Program Director, Aural/Oral Habilitation Program, Secondary School Department, Colorado School for the Deaf and Blind, Colorado Springs, Colorado

Responsibilities included working with a multi-disciplinary team of professionals to develop a Habilitation Program in the secondary Deaf School and to provide scheduling and direct service to approximately 100 children.

Curriculum Vitae

Lisa Ann Miller

Date of Birth February 28, 1962

Work Address Colorado Department of Public Health and Environment
DCEED-EE-A3
4300 Cherry Creek Drive South
Denver, Colorado 80246-1530

Education

General Preventive Medicine Residency, 1991-1993
University of Colorado Health Sciences Center
Denver, Colorado

Masters of Science in Public Health, 1991-1993
University of Colorado Health Sciences Center
Denver, Colorado

Internal Medicine Internship, 1989-1990
University of Colorado Health Sciences Center
Denver, Colorado

Doctor of Medicine, 1989
University of Minnesota
Minneapolis, Minnesota

Bachelor of Science, 1985
Major: Food Science, Concentration: Nutrition
University of Minnesota
Minneapolis, Minnesota

Work Experience

Medical Director, Colorado Responds to Children with Special Needs, Colorado Department of Public Health and Environment, 11/97-present
Denver, Colorado

Medical Epidemiologist, Colorado Department of Public Health and Environment, 11/93-11/97
Denver, Colorado

Medical Epidemiologist/Preventive Medicine Resident, University of Colorado Health Sciences Center, 3/93-8/93 Denver, Colorado
Medical Epidemiologist/Preventive Medicine Resident, Colorado Department of Health, 3/92-2/93
Denver, Colorado

Research Physician/Preventive Medicine Resident, Denver Public Health Disease Control, 9/91-2/92

Denver, Colorado

Appointments

Sr. Instructor Adjoint in the Department of Preventive Medicine and Biometrics, University of Colorado Health Sciences Center

Publications/Presentations

"The Effect of HIV Counseling/Testing on STD Incidence and Sexual Behavior in an STD Clinic" (poster) American Public Health Association Meeting, Washington D.C., 1992.

"Denver's Increase in HIV Counseling after Magic Johnson's HIV Disclosure" (letter) Cohn DL, Miller LA, Yamaguichi KJ, Douglas JM. Am J Public Health 1992;82:1692.

"Comparison of anonymous and confidential HIV test sites: Colorado, 1990-1992" Hoffman R, Cohn D, Hoxworth T, Miller L, Guilfoile A, Hamman R, et al. IXth International Conference on AIDS, Berlin, 1993.

"Risk Factors for Delayed Immunization against Measles, Mumps, and Rubella in Colorado Two-Year Olds" Miller LA, Hoffman RE, Baron AE, Marine W, Melinkovich P. Pediatrics 1994;94:213-219.

"Carbon Monoxide Poisoning - Weld County, Colorado, 1993" Cook M, Miller L, Hoffman R, Clem B, et al. Morbidity and Mortality Weekly Report 1994;43:765-767.

"Comparison of Partner Notification at Anonymous and Confidential HIV Test Sites in Colorado" Hoffman RE, Spencer NE, Miller LA. JAIDS 1995;8:406-410.

"Surveillance for fetal alcohol syndrome in Colorado" Miller LA, Shaikh T, Stanton C, Montgomery Rickard R, Keefer S, Hoffman R. Public Health Reports 1995;1 10:690-697.

“Neural tube defects surveillance: a national survey” Miller LA, Kirby RS. Teratology 2000; 1 /2: 28-32.

Curriculum Vitae

Vickie Thomson
4665 Kirkwood Court

Boulder, CO 80301
303-581-9120

Educational Background

University of Northern Colorado, Audiology MA, 1978
University of Northern Colorado, Audiology, BA, 1977

Professional Experience

Colorado Department of Public Health and Environment, Health Care Program for Children with Special Needs, Denver, CO, 1991-present

State Audiology Consultant (1998-2000). Assure comprehensive audiological services for children birth through 21 years of age; promote statewide interagency collaboration to assure community based services; develop statewide Audiology policies and procedures; provide indirect supervision to the state regional coordinators; provide in-service and training;

Colorado Infant Hearing Program Coordinator (1991- present). Provide consultation and coordination in the development of statewide activities for newborn hearing screening programs per HB 1095-97; develop data management and tracking systems for newborn hearing screening programs; assurance that a comprehensive system from screening through intervention for infants identified with hearing loss; provide technical assistance to other MCH programs through out the United States; Co-Chair the Colorado Infant Hearing Advisory Committee

Co-Principle Investigator of the CDC Early Hearing Detection and Intervention Grant (EHDI) (2000-present). Assure the goals and objectives of the Grant are meet; develop a data management and tracking system that will integrate with other CDPHE departments to provide better care coordination for families statewide; coordinate Grant activities with grant staff and other departments such as CRCSN, Vital Statistics, EPSDT, and Metabolic Genetic Disorders.

Boulder Community Hospital, Mapleton Center for Rehabilitation, Boulder, CO, 1986-1998, 2000-present

Clinical audiologist, program coordinator. Provide routine audiological evaluations, auditory brainstem response evaluations, intraoperative monitoring, electronystagmography and amplification services for pediatric through geriatric populations; develop policies and procedures to meet JCAHO requirements; supervise audiologists and newborn hearing screening coordinators; provide program development and marketing plans

University of Colorado, Boulder, CO, 1996-2000

Project Coordinator, Maternal and Child Health Grant, Marion Downs National Center for Infant Hearing. Provide technical assistance to participating grant States in the area of newborn hearing screening; provide national in-services and presentations; coordinate conference activities. Assistant professor for graduate classes in auditory brainstem response, electronystagmography, and infant evaluations

Western Otolaryngology, Denver, CO, 1979-1986

Clinical audiologist. Provide routine audiological evaluations, auditory brainstem response audiometry, electronystagmography and amplification services for pediatric through geriatric populations.

Professional Affiliations

Colorado Academy of Audiology, Vice President, 1999

Directors of Speech and Hearing Programs in State Health and Welfare Agencies, President, 1999

American Academy of Audiology, Newborn Hearing Task Force, Chair, 1997-Present
American Speech, Language and Hearing Association, Certification, 1978-Present
Colorado Audiology Registration, Department of Regulatory Agencies, exp. 6/30/99

Honors

Marion Downs National Award, 1995
Colorado Academy of Audiology Peak Performance Award, 1995
Colorado Hearing Foundation Award, 1995
Boulder Community Hospital Encore Gold Award, 1994
National Distinguished Service Registry, 1989

Publications

Mehl, A., Thomson,V. The Colorado newborn Hearing Screening Project: 1992-1999: On the Threshold of Effective Population-Based Universal Newborn Hearing Screening. Pediatrics, Vol.109.No1January2002

Thomson,V.,et.al The Marion Downs National Center for Infant Hearing: Developing Comprehensive State Systems. The Otolaryngologic Clinics of North America. Vol 32, no.6, Dec. 1999

Arehart K.H.,Yoshinaga-Itano C., Thomson V.State of the States: The Status of Universal Newborn Hearing Screening, Assessment, and Intervention Systems in 16 States. American Journal of Audiology, vol 7, no.2, 101-104, 1998.

Thomson V. The Colorado Newborn Hearing Screening Project. American Journal of Audiology, 1997.

Thomson V., Rose L., O'Neal J. Statewide Implementation of Universal Newborn Hearing Screening. Seminars in Hearing, vol.19, no.3, 287-300, 1998.

Mehl A., Thomson V. Newborn Hearing Screening: The Great Omission. Pediatrics, 101-108,1998.

Gabbard S.,Thomson V., Stredler Brown A. Considerations for Universal Newborn Hearing Screening, Audiologic Assessment, and Intervention. Audiology Today, 8-10, 1998

Mehl A., Thomson V. Universal Newborn Hearing Screening: An evolving standard of care for neonates. Audiology Today, 28-29, 1998

CURRICULUM VITAE

NAME: James Clyde Ledbetter, M.D., F.A.A.P.
DATE OF BIRTH: March 29, 1956
PLACE OF BIRTH: Bartow, Florida, USA
PERSONAL DATA: Wife: Jennifer; Children: Caroline, Sara and Grace

CURRENT HOME ADDRESS: 826 South York
Denver, Colorado 80209-4647
(303) 778-7857

PROFESSIONAL MEMBERSHIPS:
Fellow, American Academy of Pediatrics
Member, Section on Developmental and Behavioral Pediatrics, AAP
Member, Section on Children with Disabilities, AAP
Member, Society for Developmental Pediatrics
Member, Section on Emergency Medicine, AAP

CERTIFICATION:
1993 Medical Licensure - State of Colorado, current
1991 Diplomat - American Board of Pediatrics
1990 Medical Licensure - State of Ohio, current
1989 National Board of Medical Examiners

COMMUNITY OUTREACH:
Colorado Interagency Coordinating Council, appointed by the governor
September 2000 to present

Board of Directors, United Cerebral Palsy
2000 to present, secretary 2001-2002

Health Advisory Committee, Creative Options
Head Start, Aurora
2000 to present

Physician Advisory Board
Health Care Program for Children with Special Needs, Colorado
1997 to present

Member, Committee for Public Awareness
Early Childhood Connections (part C services of IDEA)
1996 to present

HOSPITAL COMMITTEES:
Emergency Service Function, Medical Center of Aurora
1999 to 2001

PROFESSIONAL EXPERIENCE:
Medical Consultant current

Health Care Program for Children with Special Needs
Dept. of Public Health and Environment, Colorado

Developmental Pediatrician for Northwest D & E Clinic, 11/93 to Present
Dept. of Public Health and Environment, Colorado

Pediatrician, Carepoint P.C. 6/98 to 12/01
Emergency Department, Medical Center Aurora
Aurora, Colorado

Independent Contractor, General & Developmental Pediatrics 1/98 to 6/98

Denver, Colorado

Developmental Pediatrician, Private Practice, 7/97 to 12/97
Colorado Neurological Services P.C.
Denver, Colorado

Assistant Professor, Pediatrics 8/95 to 6/97
University of Colorado School of Medicine
Medical Director, Special Needs Primary Care Clinic
The Children's Hospital, Denver, CO

Developmental Psychobiology Research Fellow, 11/94 to 6/95
UCHSC Denver, CO

Fellow - Developmental Pediatrics 7/93 to 6/95
University of Colorado Health Sciences Center

U.S. Air Force Medical Corps, KISAFB, MI 7/91 to 6/93
Staff Pediatrician, along with one other pediatrician, providing all-inclusive care to pediatric dependents of active duty and retired military personnel in the local area.

University of Florida College of Medicine, Gainesville, FL 6/83 to 8/84
Physician Assistant, Department of Otolaryngology
Under the supervision of four otolaryngologists, performed clinical duties in surgery, clinic and the hospital ward.

Gainesville Veterans Medical Center, Gainesville, FL 3/82 to 3/83
Physician Assistant, Division of Thoracic Surgery
Responsible for orchestrating and providing pre and post-operative care of veterans requiring thoracic surgery.

Florida Health Care HMO, Daytona Beach, FL 2/81 to 2/82
Physician Assistant
Provided care to HMO members in the Urgent Care Clinic under the indirect supervision of HMO physicians.

Halifax Medical Center, Daytona Beach, FL 9/80 to 2/81
Surgical Assistant
First assistant in vascular, orthopedic, plastic, general and OB/GYN,
and second assistant in neurosurgery.

EDUCATION:

1994-1995	Research Fellowship Developmental Psychobiology Research Group University of Colorado Health Science Center Denver, Colorado
1993-1995	Fellowship, Developmental Pediatrics J.F. Kennedy Center for Developmental Disabilities University of Colorado Health Science Center Denver, Colorado
1988-1991	Pediatric Residency Children's Hospital Medical Center Cincinnati, Ohio
1984-1988	Medical School Degree: M.D. University of Florida College of Medicine Gainesville, Florida
1978-1980	Undergraduate Degree: B.S. in Medicine Physician Assistant Program University of Florida Gainesville, Florida
1974-1976	Undergraduate (no degree) U.S. Naval Academy Annapolis, Maryland

AWARDS AND ACHIEVEMENTS OF NOTE:
Lecturer, Edward Pratt Lecture Series, 1991, Children’s Hospital Medical Center, Cincinnati, OH.
Chief Residents' Award, 1991, Children’s Hospital Medical Center, Cincinnati, OH.
Facilitator of Peer Learning from the class of 1988, College of Medicine, U of Florida.
Florida Pediatric Society Award for the class of 1988, College of Medicine, U of Florida.
Genevra Todd and Henry Meleny Award for class of 1988, College of Medicine, U of Florida.

AREAS OF INTEREST:
Autism, medical conditions associated with developmental disabilities, pharmacology of maladaptive behaviors, the medical home for children with special health care needs.

APPENDIX L: COMMITTEE MEMBERSHIP

Colorado's Newborn Metabolic [Bloodspot] Screening Program Advisory Committee
February, 2001

(Each of the below listed entities has one voting member of the Committee, the state representatives are non-voting)

American Academy of Pediatrics, Colorado Chapter
Colorado Academy of Family Physicians
Colorado Health and Hospital Association
Colorado Perinatal Care Council
Colorado Rural Health Center
Colorado Society of Clinical Pathologists
Low-Risk Neonatal Nurses of Colorado
Cystic Fibrosis Center @ Denver Children's Hospital

Inherited Metabolic Disease Clinic @ Denver Children's Hospital

Pediatric Endocrinology @ Denver Children's Hospital
@ Private Pediatric Endocrinology Office in Englewood, Colorado

Sickle Cell Research and Treatment Center @University of Colorado Health Sciences Center

DNA Diagnostic Laboratory @University of Colorado Health Sciences Center

Consumer
Consumer

<u>Colorado Department of Public Health and Environment Staff Members of the Committee (non-voting members)</u>
Chief Microbiologist, State Laboratory
Colorado Responds to Children with Special Needs (state birth defects registry), Medical Director
MCH Medical Consultant/Director of Newborn Screening Follow-up Program
Newborn Screening Follow-up Program Coordinator
Newborn Screening Laboratory Supervisor
Newborn Screening Laboratory Senior Chemist
State Genetics Coordinator

This committee reports to the Director of the Colorado Department of Public Health and Environment's Laboratory and Radiation Services Division and to the Director of the Division of Prevention and Intervention Services for Children and Youth (which includes all Title V programs)

Of the fourteen voting membership categories, two are for consumers.

STATE GENETICS ADVISORY COMMITTEE
October 2000

Mr. John Astuno
1775 Sherman St., Ste. 1650
Denver, CO 80203
email: normaCOB@aol.com
phone: 303-861-7636
fax: 303-832-1405

Ms. Susan Bryan, Contractor
Quantum Trust
7538 N. Sixth Place
Phoenix, AZ 85020
email: sbryan@quantumtrust.com
phone: 602-870-4752
fax: 602-870-4782

Dr. Tim Byers
UCHSC
Dept. of Preventive Medicine & Biometrics
4200 E. 9th Ave., Box C-245
Denver, CO 80262
email: tim.byers@uchsc.edu
phone: 303-315-5169
fax: 303-315-3183

Ms. Judy Capra
4290 Teller St.
Wheat Ridge, CO 80033
email: jcapra@jeffco.k12.co.us
phone: 303-467-1184
fax: 303-982-2834

Dr. Marilyn Coors
UCHSC
4200 E. 9th Ave., Box B-137
Denver, CO 80262
email: marilyn.coors@uchsc.edu
phone: 303-315-0203 fax: 303-393-7798

Dr. Richard Jones
Mountain States Maternal-Fetal Med.
2005 Franklin St, Midtown 1, Ste. 750
Denver, CO 80205
email: rojmfm@aol.com
phone: 303-837-7885
fax: 303-837-7967

Ms. Donna Dixon
Colorado Sickle Cell Treatment/
Research Center - UCHSC
4200 E. 9th Ave., Box C-222
Denver, CO 80262
email: donna.dixon@uchsc.edu
phone: 303-372-9070
fax: 303-372-9161

Ms. Cindy Ehnes
Commission on Family Medicine
1180 Clermont St.
Denver, CO 80220
email: cindy.ehnes@uchsc.edu
phone: 303-315-9707
fax: 303-315-9752

Ms. Heidi Frey, J.D.
Patient Advocacy Coalition
850 E. Harvard Ave., Ste. 465
Denver, CO 80210
email: hfrey@patientadvocacy.net
phone: 303-744-7667
fax: 303-744-7876

Ms. Kathy Gaines
Director of Program Services
Colorado March of Dimes
1325 S. Colorado Blvd., Ste. 508
Denver, CO 80220
email: kgaines@modimes.org
phone: 303-692-0011
fax: 303-692-9229

Dr. Mark Johnson
Executive Director
Jefferson County Dept. of Health and Env.
1801 E. 19th St.
Golden, CO 80401
email: mjohnson@co.jefferson.co.us
phone: 303-271-5700

Dr. David Patterson
Eleanor Roosevelt Institute
1899 Gaylord St.
Denver, CO 80206
email: davepatt@eri.uchsc.edu
phone: 303-336-5650
fax: 303-333-8423

Dr. Peter Lane
Colorado Sickle Cell Treatment/
Research Center
UCHSC
4200 E. 9th Ave., Box C-222
Denver, CO 80262
email: peter.lane@uchsc.edu
phone: 303-372-9070
fax: 303-372-9161

Dr. David Manchester
The Children's Hospital
Dept. of Genetics, B-300
1056 E. 19th Ave.
Denver, CO 80218
email: manchester.david@tchden.org
phone: 303-861-6395
fax: 303-861-3921

Dr. Loris McGavran
Director
Colorado Genetics Laboratory
4200 E. 9th Ave., Box C-225
Denver, CO 80262
email: loris.mcgavran@uchsc.edu
phone: 303-315-7249
fax: 303-315-7044

Dr. Stefan T. Mokrohisky
Kaiser Permanente
Department of Pediatrics
1375 E. 20th Ave.
Denver, CO 80205
email: stefan.mokrohisky@uchsc.edu
phone: 303-861-3558
fax: 303-861-3667

Matthew R.G. Taylor, M.D.
Division of Genetics
The Children=s Hospital
1056 E. 19th Ave., B-300
Denver, CO 80218
email: matthew.taylor@uchsc.edu
phone: 303-837-2853
fax: 303-861-3921

Ms. Carol Walton, M.S.
The Children=s Hospital
Division of Genetics, B-300
1056 E. 19th Ave.
Denver, CO 80218
email: walton.carol@tchden.org
phone: 303-861-6839

Ms. Cindy Quince, Staff
9406 Desert Willow Trail
Highlands Ranch, CO 80126
email: acquince@att.net
phone: 303-471-5101
fax: (currently unavailable)

Mr. Barry Rosenberg
Rocky Mountain Chapter
178 Parkview Ave.
Golden, CO 80401
email: boldtzah@aol.com
phone: 303-233-3122
fax: 303-233-1478

Ms. Bessie Smith
2839 Decatur St.
Denver, CO 80211
email: bessie.smith@state.co.us
phone: 303-692-2433
fax: 303-782-5576

Dr. Richard Spritz
Professor and Director
Human Medical Genetics Program
UCHSC
4200 E. 9th Ave., Box B-161
Denver, CO 80262
email: richard.spritz@uchsc.edu
phone: 303-315-0409
fax: 303-315-0407

Ms. Ann McNulty
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
FCHSD-DAT-A4
Denver, CO 80246
email: ann.mcnulty@state.co.us
phone: 303-692-2311
fax: 303-782-5576

Lisa Miller, M.D.
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
DCEED-DS-A3
Denver, CO 80246
email: lisa.miller@state.co.us
phone: 303-692-2663

fax: 303-861-3921

Dr. Mark Yarborough
UCHSC
4200 E. 9th Ave., Box B-137
Denver, CO 80262
email: mark.yarborough@uchsc.edu
phone: 303-315-5177
fax: 303-393-7798

Ms. Joyce Hooker
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
FCHSD-MAS-A4
Denver, CO 80246
email: joyce.hooker@state.co.us
phone: 303-692-2423
fax: 303-782-5576

Dr. Bill Letson
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
FCHSD-MAS-A4
Denver, CO 80246
email: bill.letson@state.co.us
phone: 303-692-2424
fax: 303-782-5576

fax: 303-782-0904

Normie Morin, Ph.D., M.P.H.
Chief, Chronic Disease Section
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
EMSPD-A5
Denver, CO 80246
email: normie.morin@state.co.us
phone: 303-692-2505
fax: 303-782B0095

Ms. Kathy Watters
CO Dept. of Public Hlth & Env.
4300 Cherry Creek Drive South
FCHSD-HCP-A4
Denver, CO 80246
email: kathy.watters@state.co.us
phone: 303-692-2418
fax: 303-782-5576

Medical Home Advisory Committee

LYNN BINDEL RN MS CNS
HCP NURSING CONSULTANT

CHRISTIE BLAKELY
69 SPYGLASS
LITTLETON CO 80123

DON COOK MD
1710 21ST AVENUE
GREELEY CO 80631

JOAN EDEN MSPH RD
MCH ACTING DIRECTOR

DEBBIE EFIRD MD
1583 S EMERSON
DENVER CO 80210

LISA M ESGAR RN MSN
DIV OF HEALTH PLAN MGMT
1575 SHERMAN ST 5TH FLOOR
DENVER CO 80203-1714

ED GOLDSON MD
THE CHILDRENS HOSPITAL
1056 E 19TH AVE B140
DENVER CO 80218

VIRGINIA GURLEY MD
501 S CHERRY ST SUITE 700
DENVER CO 80246

ELIZABETH SOPER HEPP
COLO DEPT OF SPECIAL ED
201 E COLFAX
DENVER CO 80203

PAULA HUDSON PHD CCC
HCP SPEECH/LANG CONSULT

SANDRA JACQUEZ
HCP PROVIDER RELATIONS

JIM LEDBETTER MD
826 S YORK
DENVER CO 80209

PAUL MELINKOVICH MD
DENVER HEALTH MED CTR
660 BANNOCK MC 1914
DENVER CO 80204

JENNIFER MILES
COLO COMM HEALTH NETWORK
800 GRANT ST SUITE 505
DENVER CO 80203

LORRI PARK
1843 SPRUCE
BOULDER CO 80302

CAROL REIDER
HCPF
1575 SHERMAN
DENVER CO 80203

CORRY ROBINSON PHD
JFK PARTNERS
1825 MARION ST
DENVER CO 80218

PATTI ROSEQUIST MD
3260 15TH ST
BOULDER CO 80304

MARK WALLACE MD
WELD CNTY HEALTH DEPT
1555 N 17TH AVENUE
GREELEY CO 80631

KATHY WATTERS MA CCC
HCP ACTING DIRECTOR

MIKE YOUNG MD
MEDICAL DIRECTOR CHPR
400 S COLORADO BLVD STE 300
DENVER CO 80222

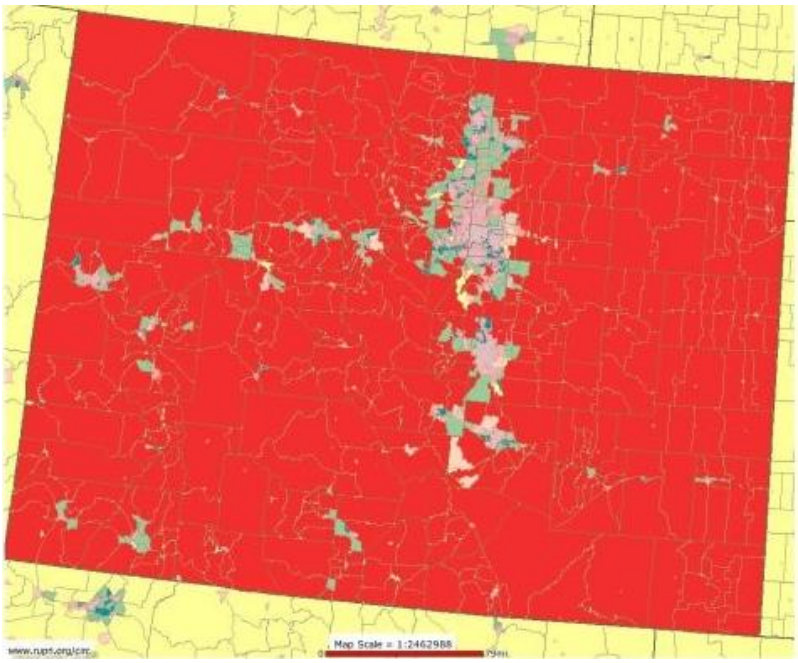
DAVID HERR MD
ROCKY MOUNTAIN HMO
2775 CROSSROADS BLVD
PO BOX 10600
GRAND JUNCTION CO 81502-5600

LINDA THERRIEN
110 16TH ST SUITE 1000
DENVER CO 80202

APPENDIX M: ORGANIZATIONAL CHARTS

APPENDIX N: FRONTIER, RURAL AND URBAN COUNTIES

Rural and frontier areas appear in red



APPENDIX O: IRB PROTOCOL FOR EHDI PROJECT

(contains detailed description of EHDI Project, Purposes, Processes and description of IRIS as it relates to EHDI).

APPENDIX P: IRB SUMMARY REQUIREMENTS

Colorado Department of Public Health and Environment

Title of Project: Long-term Multiple Database Integration and Follow-up of Infants with Special Health Care Needs

Time Period Covered by Project: Indefinite although grant funding is for 5 years beginning September 1, 2000.

Principal Investigator: Vicky Thomson MA, Bill Letson MD and Lisa Miller MD

Project Director: Kathy Watters MA **Telephone Number:** 303-692-2424

Specify Grant, Contract, Fellowship, Cooperative Agreement or Other: Centers for Disease Control and Prevention grant entitled "Early Hearing Detection Intervention System and Newborn Screening Data Integration Program."
Funding Source: Centers for Disease Control and Prevention
Grant Number No: UR3/CCU818868-01
Program Announcement: 00076

Purpose of Study: The above named grant is intended to integrate various databases into a common database, with the primary concerns being 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) that manages client information from the Early Periodic Screening, Diagnosis and Treatment program and the Health Care Program for Children with Special Needs (HCP). IRIS is currently undergoing a revision that will make it capable of case management functions by allowing the system to contain a limited electronic medical record.

There are three primary purposes in integrating these databases into IRIS:

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. This will make 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.

How will subjects be recruited? Subjects will be identified during their regular contacts with the health care system. Every child with a specific confirmed diagnosis will be eligible for this long-term follow-up. Such a diagnosis will follow a positive screen in the NBMS or NBHS system. Families will be asked for consent by the specialty providers involved in the clinical care of the infant's specifically diagnosed condition(s) or by their primary care providers. If children with specified diagnoses are not in the care of appropriate specialists and their primary care providers are not known, their families will be contacted by their County Public Health Nurses. If the families consent, the follow-up process would then begin. In Denver County, which has no Public Health Nurses, contact will be made with the families by the Children and Families Program at Denver Health Medical Center. This program is a special program oriented to providing case management for Denver County children with special health care needs.

What will subjects be asked to do? At present, CDPHE has personnel dedicated to assuring appropriate confirmatory testing for infants who have tested positive on the NBMS and NBHS screens. These screens are performed twice in the child's first two weeks of life. When a positive NBMS result is determined by the CDPHE laboratory, the physician of record or other submitter is notified by the lab and the abnormal screening result is passed on to the follow-up unit in the Prevention and Intervention Services for Children and Youth Division (PSD) of CDPHE. In the case of NBHS, the birthing hospitals notify CDPHE of the screening results. In both screening systems, the CDPHE follow-up personnel then pursue the case of a positive screen until it is determined that the child is known to a responsible primary medical provider who can refer the child for appropriate confirmatory testing and sub-specialty consultation if need be. Primary medical providers are defined as physicians, physician assistants or nurse practitioners that provide primary health care to the child in question.

Our purpose is to foster a more comprehensive follow-up process that, with the assistance of electronic databases, will foster necessary follow-up for the infants, assist health care providers in obtaining critical information about these infants, and facilitate the disease monitoring purposes identified above. To accomplish this, parents of diagnosed children will be asked to: 1) Allow information about their child to be shared among providers with certain specified restrictions 2) agree to contact by local Public Health Nurses and 3) agree to have information on their children in the integrated database.

Risks of this project: The risk of this project is the very small risk that information pertaining to a given child would be seen by someone who is not authorized to see the information and that person would somehow misuse the information. The security procedures used for this system, including parent control of access to their child's information by using a key in conjunction with a provider key, are described below under "How will confidentiality of data be ensured?" These procedures will make the risk of the wrong people seeing the information very small. There is, then, an even smaller but real risk that the wrong people seeing the information would misuse it in some way.

Benefits of the project: The benefit to a family in having a child's information in the electronic database is the ability to very quickly transfer critical information to new health care providers the child may come in contact with. It will also help to assure that a child is getting the best of what health care is available in the area the family lives and to help arrange for necessary care when care is not available locally.

What types of data will be collected on subjects? The information on all the forms (Appendices B-C2) will be in ACCESS or similar databases that can be downloaded into IRIS at CDPHE, a case management database for children with special health care needs. At this point we have designed data collection modules for Sickle Cell Disease, Inherited Metabolic Diseases, and congenital hearing loss. We anticipate developing similar modules for congenital hypothyroidism, congenital adrenal hyperplasia and cystic fibrosis in the next year.

When circumstances necessitate their involvement, Public Health Nurses will follow the children on an annual basis at minimum, to determine if the child has a primary care provider and if there have been necessary referrals to appropriate specialty care providers. Public Health Nurses would have access only to information needed to allow their tracking and follow-up. This information will be limited to the information in Appendix B unless express written parental permission is given for access to more detailed information in Appendices C1 and C2. This process will involve an exchange of information between medical providers, local Public Health Nurses and the state surveillance system in cases where assurance is needed that adequate follow-up and treatment is being rendered to a child with a diagnosed condition.

The type of information in Appendices B-C2 is already collected by specialty providers on significant aspects of the children's' clinical care and will be used, in aggregate, to determine long term outcome. It will also provide the ability to analyze risk factors for poor outcomes. Specialty medical providers include physician specialists, physician sub-specialists and audiologists who provide complex specific specialty care to the child in question. The information in Appendices B-C2 would also be available on a need to know basis to appropriate primary and other specialty providers as part of the patients' clinical care management. These-additional providers would have to have written parental consent in order for them to receive an electronic copy of any information in IRIS that they themselves had not entered.

How will data be collected and analyzed? The primary data collection point will be the specialists' clinical practice for clinical and most annual follow-up information. For children who do not have appropriate specialty care, Public Health Nurses will collect primary data on the basic case management and follow-up information (Appendix B). As part of their case management, these nurses will work at coordinating or facilitating appropriate primary and specialty care for the child in question. The flow of follow-up information is shown in Appendix A and the database will serve as the repository for case management documentation. This basic follow-up documentation can then be available to health care providers involved in a child's care.

Data will be analyzed to determine completeness of case identification and follow-up by checking consistency between differing primary datasets. For example, Colorado's birth defects registry, Colorado Responds to Children with Special Needs (CRCSN) should contain all of the children with a specific diagnosis from the NBMS and NBHS screening. By linking data in IRIS with CRCSN data we will be able to test how complete the reporting and collecting of information is in one primary data system compared to the other primary data system where children should be entered in both. It will also help to determine the efficiency and effectiveness of the electronic data flow. By linking these datasets with birth certificate data, it will be possible to identify risk factors for poor follow-up and poor outcomes through statistical analysis of the linked datasets. This will, in turn, allow assessment of those risk factors that are potentially modifiable. Then, in conjunction with specialty clinical providers, we will be able to design interventions aimed at altering the modifiable risk factors so that long-term

outcomes for children in the database might be improved. The efficacy of specific clinical practices could also be evaluated in some circumstances by analysis of the data collected on Appendices B-C2.

How will confidentiality of data be ensured? Access to provider databases will be limited to those with a need to know and protected by passwords. Access to information in IRIS pertaining to specific children by information seekers outside CDPHE will be through a dual keyed system: An identifying key from the mother (such as a child's social security number) and a provider specific number, such as a medical license number. This will provide access only to a specific child's data by consented agreement of the parent as in Appendix D and by the parent's giving their child's access number to the provider at hand. Procedures for provider enrollment and agreement to confidentiality procedures are in Appendix E. The scope of information that could be accessed by health providers would be limited in a couple of key ways: 1) Access would be limited to information that the provider had themselves provided to the data-base and 2) would include basic information on the children that is important for immediate public health purposes such as NBMS and NBHS results and immunization status (once the recent legislatively authorized immunization tracking system is functional and available to IRIS). While the outside provider could not access the data provided by the sub-specialist, contact information for the sub-specialist would be available. Further access to sub-specialty records would be granted through the standard process used currently for paper medical records, i.e. signed parental permission for copy and transfer of paper records or transfer of electronic records to the provider in question.

Access to the IRIS database by CDPHE staff will be limited to those program staff actively involved in the initial screening follow-up or data collection; individuals who need specific access for the purpose of analysis of data (including linkage to other datasets); and to local Public Health Nurses in the County of the child's residence in cases where adequate follow-up is not being accomplished by appropriate specialists. If a child should move to a different county, access rights will be made available to PHNs in the new county of residence. Individual permission for partial access to the database at CDPHE by state or county employees will be obtained under established procedures for access to vital statistics data**.** CDPHE staff confidentiality agreements and procedures are in Appendix F.

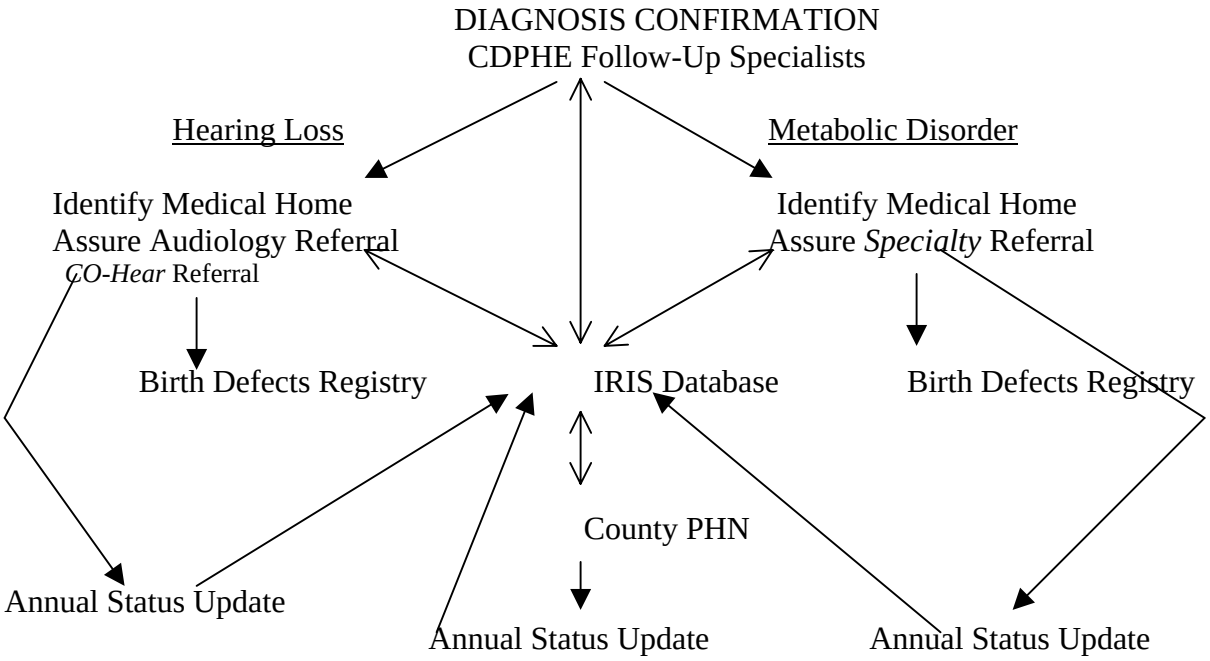
Who will have access to the data and how long will the data be kept? Only CDPHE program and administrative staff (including Public Health Nurses acting as program staff and having signed the CDPHE confidentiality agreement) and providers who have signed an agreement to follow program procedures will have access to the data. The data are to be kept indefinitely for the purpose of long-term follow-up and assessment of risk for poor outcome or poor follow-up. The data is electronically secured and encrypted as described in Appendix G.

How will the data be presented in a final report? This is a long-term project with periodic reporting. The data will be reported periodically, in aggregate and without identifiers, and in a way that is consistent with CDPHE Policy on Release of Disease Surveillance Data.

Include a copy of the consent form following the consent form guidelines. See Appendix D.

APPENDIX Q: FOLLOW-UP SCHEMATIC FOR INFANTS

FOLLOW-UP SCHEMATIC FOR INFANTS DIAGNOSED WITH CONDITIONS INITIALLY DETECTED BY NEWBORN SCREENING



NEWBORN SCREENING FOLLOW-UP FORM (Appendix B)
Last update entry: MM/DD/YY

	Current Last	Previous Last	First	Middle
Child's Name:	_____	_____	_____	_____
Mother's Name:	_____	_____	_____	_____
Father's Name:	_____	_____	_____	_____

Identification #: NBS lab #:---------- Electronic Birth Certificate # ----------

Date of Birth: MM/DD/YY *Gender*: M F

Ethnicity: Caucasian__ African-American__ Asian/Pacific Islander__ Hispanic__
American Indian/Alaska Native__ Other__ Unknown__

1st metabolic screen date: MM/DD/YY Normal__ Abnormal__
2nd metabolic screen date: MM/DD/YY Normal__ Abnormal__

1st hearing screen date: MM/DD/YY Normal__ Abnormal__
2nd hearing screen date: MM/DD/YY Normal__ Abnormal__

Diagnosis (Dx): 1)_____ *Date of Dx confirmation*: 1) MM/DD/YY
Diagnosis (Dx): 2)_____ *Date of Dx confirmation*: 2) MM/DD/YY
Diagnosis (Dx): 3)_____ *Date of Dx confirmation*: 3) MM/DD/YY

(have space for diagnosis name and corresponding ICD-9 code)

First Colorado Primary Provider_____ Phone #: --- --- ----

First Colorado Specialty Provider_____ Phone #: --- --- ----

Status: Deceased Y N Date of Death: MM/DD/YY Cause of Death:_____

In State? Y N Unk In Care? Y N Unk

Moved To:_____ Date: MM/DD/YY
Transfer of Care Confirmed? Y N Unk New Provider Name_____

Medical Home/Primary Provider_____ Phone #: --- --- ----

Specialty Provider_____ Phone #: --- --- ----

HEMOGLOBINOPATHY MODULE (Appendix C-1)

Last update entry: MM/DD/YY

	Current Last	Previous Last	First	Middle
Child's Name:	_____	_____	_____	_____

Mother's Name: _____ _____ _____ _____
Father's Name: _____ _____ _____ _____

Identification #: NBS lab #:---------- Electronic Birth Certificate # ----------

Date of Birth: MM/DD/YY *Gender*: M F

Diagnostic Test Result:_____ Date: MM/DD/YY

Diagnosis (Dx):_____ *Dx Confirmation Date*: MM/DD/YY

Referred to Sickle Cell Center? Y N Date of 1st visit: MM/DD/YY

*Current Primary Provider*_____ Phone #: --- --- ----

*Current Hematologist*_____ Phone #: --- --- ----

Penicillin Prophyllaxis started? Y N Unk NA Date of Start: MM/DD/YY
Penicillin Prophyllaxis stopped? Y N Unk NA Date of Stop: MM/DD/YY

Dates of Pneumoccal Conjugate Vaccination: 1) MM/DD/YY 2) MM/DD/YY
3) MM/DD/YY 4) MM/DD/YY 5) MM/DD/YY

Dates of Pneumococcal Polysaccharide Vaccination: 1) MM/DD/YY 2) MM/DD/YY

Dates of Hib Conjugate Vaccination: 1) MM/DD/YY 2) MM/DD/YY 3) MM/DD/YY
4) MM/DD/YY 5) MM/DD/YY

Dates of Sickle Cell Center Health Maintenance: 1) MM/DD/YY 2) MM/DD/YY
3) MM/DD/YY 4) MM/DD/YY 5) MM/DD/YY 6) MM/DD/YY 7) MM/DD/YY

Last Date of Health Status Review? MM/DD/YY

Clinical Complications:

Chest Syndrome: Date: MM/DD/YY Hospitalized? Y N **(4/yr)**
Admission Date: MM/DD/YY Discharge Date: MM/DD/YY
Intubated? Y N Simple Transfusion? Y N Exchange Transfusion? Y N

Splenic Sequestration: Date: MM/DD/YY Transfusion? Y N Hospitalized? Y N
Admission Date: MM/DD/YY Discharge Date: MM/DD/YY **(3/yr)**

Aplastic Crisis: Date: MM/DD/YY Transfusion? Y N Hospitalized? Y N
Admission Date: MM/DD/YY Discharge Date: MM/DD/YY **(2/yr)**

Stroke: Date: MM/DD/YY Hospitalized? Y N **(2/yr)**
Admission Date: MM/DD/YY Discharge Date: MM/DD/YY

Priapism > 3 hours: Date: MM/DD/YY Irrigated? Y N **(6/yr)**
Hospitalized? Y N Admission Date: MM/DD/YY Discharge Date: MM/DD/YY

Pain presenting to medical provider: Date: MM/DD/YY **(12/yr)** Hospitalized?
Y N Admission Date: MM/DD/YY Discharge Date: MM/DD/YY

Bacteremia: Date: MM/DD/YY Hospitalized? Y N **(3/yr)**
Admission Date: MM/DD/YY Discharge Date: MM/DD/YY
ICU? Y N Central Line? Y N
Organism: _____ _____ Antibiotic Sensitivities_____

Chronic Transfusions? Y N Date of Start: MM/DD/YY **(2/yr)**
Date of Stop: MM/DD/YY

Hydroxyurea? Y N Date of Start: MM/DD/YY **(2/yr)**
Date of Stop: MM/DD/YY

Bone Marrow Transplant? Y N Date: MM/DD/YY
Outcome: 1) Alive/well 2) graft failure 3) chronic tx complications

INHERITED METABOLIC DISEASE MODULE (Appendix C-2)

Last update entry: MM/DD/YY

	Current Last	Previous Last	First	Middle
Child's Name:	_____	_____	_____	_____
Mother's Name:	_____	_____	_____	_____
Father's Name:	_____	_____	_____	_____

Identification #: NBS lab #:---------- Electronic Birth Certificate # ----------

Date of Birth: MM/DD/YY *Gender*: M F

<u>GENERAL DATA:</u> Diagnosis_____

Family History: Y N Affected Parents Y N Names_____

Affected Siblings Y N Names_____

Dates of IMD clinic Health Maintenance: 1) MM/DD/YY 2) MM/DD/YY

3) MM/DD/YY 4) MM/DD/YY 5) MM/DD/YY 6) MM/DD/YY

School Performance: Regular Education? Y N NA (not applicable)
Grade __ Age Appropriate? Y N NA

Achievement: Above Average Y N NA Average Y N NA
Below Average Y N NA

Special Education? Y N NA Full time? Y N NA Part time? Y N NA
Mainstreamed/Individual Help Y N NA
ADHD (confirmed Dx)? Y N NA
Learning Disability (confirmed Dx)? Y N NA

Clinical Developmental Assessment: Motor: Normal__ Abnormal__
Language: Normal__ Abnormal__
Neurologic symptoms? Y N Symptoms_____

Behavioral concerns? Y N If so, what? _____

SPECIFIC DATA PKU: Diagnostic phenylalanine (phe) level---.-- Date: MM/DD/YY

6 month average phe level---.--, range ---.-- to ---.-- , #-- Age in years --.-

Genotype_____

On Diet? Y N Start Date: MM/DD/YY

Formula? Y N Phenylalanine restriction? Y N Protein restriction? Y N

SPECIFIC DATA BIOTINIDASE DEFICIENCY: Diagnostic Biotinidase Activity (%)--.--

Complete Deficiency? Y N Partial Deficiency? Y N Genotype_____

Abnormal Urine Organics at Diagnosis (Dx)? Y N Hearing Loss? Y N

Symptoms at Diagnosis? Y N Seizures at Dx? Y N Alopecia/Rash at Dx? Y N

Biotin supplementation? Y N Start date: MM/DD/YY

SPECIFIC DATA GALACTOSEMIA: Diagnostic gal-1-PUT (% activity) --.-

1 year average gal-1-P --.--, range --.-- to --.--, Age in years --.-
(typically, we only do levels every three months)

Isoelectric focusing/banding_____
Patient genotype_____ Parental genotypes? Y N Father_____ Mother_____

Symptoms at Diagnosis: Liver Dysfunction? Y N Shock? Y N

Sepsis? Y N Date: MM/DD/YY Organism_____ _____

Hyperbilirubinemia: Y N Highest bilirubin level: --.- mg/dl Date: MM/DD/YY
Exchange Transfusion? Y N

Speech Disorder? Y N Cataracts? Y N Cataracts Progressive? Y N
Ovarian Dysfunction? Y N Menarche --.- Menopause --.- (both age in years)

On diet? Y N Start date: MM/DD/YY

APPENDIX R: CONFIDENTIALITY AGREEMENT

CONFIDENTIALITY AGREEMENT

The agreements made here will supersede all prior agreements, verbal or written, or those established by convention.

Confidentiality is a general standard of professional conduct. It obliges all signatories with access to personal identifying information on reportable diseases and conditions* not to discuss information with or provide copies of reports about a client, regardless of how or where acquired, to family members, friends, professional colleagues, other employees, other clients, or any other person unless such person has been authorized to have access to that information or if the information is being shared among health care providers for the purpose of providing clinical care of the individual in question.

A breach of confidentiality is defined as the release of personal identifying information (e.g., name, address, date of birth, telephone number, social security number, information that could reasonably lead to personal identification) and either:

1. other personal information (e.g., sexual orientation, drug use, etc.);
   or
2. diagnosis, test results, or the fact that a test has been performed;

to any person who does not "need to know" such information or to any other person, unless the release is necessary for the treatment, control, investigation, prevention or enforcement by public health officials. For all reportable conditions, other than AIDS or HIV infection, information is provided on the subject of the report after the subject (case) has provided CDPHE written authorization for the release for that information.

Signatories suspected of breaching confidentiality will be relieved of all duties requiring the "need to know" until an investigation of the matter is completed. A committee, consisting of the Division Director, the employee's supervisor, the State Epidemiologist, and any other persons designated by the Division Director, will listen to the involved parties, review the facts of the case, and make a determination as to whether there is evidence of a breach of confidentiality. The signatory will be given an opportunity to present his/her side of the incident to the committee. If the committee determines there is no evidence or insufficient evidence of a breach of confidentiality, the signatory will immediately have his/her "need to know" status reinstated. If the committee determines there is evidence of a breach of confidentiality, the signatory will be permanently relieved of "need to know" duties. The signatory will also be subject to the criminal penalties outlined within the federal regulations that are pursuant to the Health Insurance Portability and Protection Act of 1996.

---

* Reportable diseases and conditions are specified by regulations by the Board of Health, CDH.

The confidentiality of laboratory and case reports of reportable diseases and conditions is also protected by state law. Penalties for violating confidentiality of HIV and AIDS reports and all other reportable diseases and conditions are specified in CRS 25-4-1409 (2), CRS 25-1-122 (6) and CRS 18-1-106 (1). Upon conviction, the signatory shall be punished by a fine of not less than five hundred dollars nor more than five thousand dollars, or by imprisonment in the county jail for not less than six months nor more than twenty-four months, or by both such fine and imprisonment. It is possible that the District Attorney may prosecute an unauthorized release of confidential information as a felony offense.

I understand that medical and epidemiologic information including the names of individuals tested or reported with any disease or condition that is required to be reported by the Colorado Board of Health is confidential information. I agree that I will not reveal such confidential information, regardless of how or where acquired, to family members, friends, professional colleagues, other employees, other clients, or any other person unless such person has been authorized to have access to that information.

I further understand this agreement shall continue to bind me after I no longer have medical responsibility for any patient in question, and that unauthorized use or disclosure of any confidential information is a breach of the terms of this confidentiality agreement that allows my access to the Colorado Child Health Profile database.

I have read and understand the above information.

DATE:_____ NAME:_____

(Please Print) (Signature)

DATE:_____ WITNESS:_____

(Please Print) (Signature)

APPENDIX S: CONFIDENTIALITY AGREEMENT CDPHE

CONFIDENTIALITY AGREEMENT

The agreements made here will supersede all prior agreements, verbal or written, or those established by convention.

Confidentiality is a general standard of professional conduct. It obliges all employees and contractors of the Colorado Department of Public Health and Environment (CDPHE) with access to personal identifying information on reportable diseases and conditions* not to discuss information with or provide copies of reports about a client, regardless of how or where acquired, to family members, friends, professional colleagues, other employees, other clients, or any other person unless such person has been authorized to have access to that information.

Employees of the CDPHE Disease Control and Environmental Epidemiology Division (DCEED) who handle, access or manage laboratory and case reports of reportable diseases and conditions that contain personal identifying information have a "need to know" such information in order to perform their jobs. This confidentiality agreement identifies a person as having the "need to know" and is to be included in the employee's personnel file. A list of individuals with "need to know" status is kept on file with the Division Director.

A breach of confidentiality is defined as the release of personal identifying information (e.g., name, address, date of birth, telephone number, social security number, information that could reasonably lead to personal identification) and either:

1. other personal information (e.g., sexual orientation, drug use, etc.);
   or
2. diagnosis, test results, or the fact that a test has been performed;

to any person who does not "need to know" such information or to any other person, unless the release is necessary for the treatment, control, investigation, prevention or enforcement by public health officials. For all reportable conditions, other than AIDS or HIV infection, information is provided on the subject of the report after the subject (case) has provided CDPHE written authorization for the release for that information.

Employees suspected of breaching confidentiality will be relieved of all duties requiring the "need to know" until an investigation of the matter is completed. A committee, consisting of the Division Director, the employee's supervisor, the State Epidemiologist, and any other persons designated by the Division Director, will listen to the involved parties, review the facts of the case, and make a determination as to whether there is evidence of a breach of confidentiality. The employee will be given an opportunity to present his/her side of the incident to the committee. If the committee

* Reportable diseases and conditions are specified by regulations by the Board of Health, CDH.

determines there is no evidence or insufficient evidence of a breach of confidentiality, the employee will immediately have his/her "need to know" status reinstated. If the committee determines there is evidence of a breach of confidentiality, the employee will be permanently relieved of "need to know" duties, and other recommendations or personnel actions, including corrective action and/or disciplinary action, with possible termination and/or criminal prosecution (defined in Chapter 8 of the State Personnel Rules), may be initiated.

The confidentiality of laboratory and case reports of reportable diseases and conditions is protected by state law. Penalties for violating confidentiality of HIV and AIDS reports and all other reportable diseases and conditions are specified in CRS 25-4-1409 (2), CRS 25-1-122 (6) and CRS 18-1-106 (1). Upon conviction, the employee shall be punished by a fine of not less than five hundred dollars nor more than five thousand dollars, or by imprisonment in the county jail for not less than six months nor more than twenty-four months, or by both such fine and imprisonment. It is possible that the District Attorney may prosecute an unauthorized release of confidential information as a felony offense.

I understand that medical and epidemiologic information including the names of individuals tested or reported with any disease or condition that is required to be reported by the Colorado Board of Health is confidential information. I agree that I will not reveal such confidential information, regardless of how or where acquired, to family members, friends, professional colleagues, other employees, other clients, or any other person unless such person has been authorized to have access to that information.

I further understand this agreement shall continue to bind me during any employment within the CDPHE and even after my employment with the CDPHE has terminated, and that unauthorized use or disclosure of any confidential information is a breach of the terms of my employment with CDPHE and may subject me to court action by any interested party or to other sanctions by CDPHE. I understand it is my supervisor's responsibility to provide me with an updated list of all reportable diseases and conditions.

I have read and understand the above information.

DATE:_____ NAME:_____
(Please Print) (Signature)

DATE:_____ WITNESS:_____

(Please Print) (Signature)

cc: Employee

Division Director
Employee's Division Personnel File

APPENDIX T: LETTER

PREVENTION AND INTERVENTION SERVICES FOR CHILDREN AND YOUTH
COLORADO DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

To: Bill Letson
Fr: Ann McNulty, x2311
Re: IRIS Security

Bill, attached is a write-up about IRIS security that was submitted to Health Care Policy and Financing, for submission to HCFA.. The format is HCFA's and represents what they wanted to know about Medicaid projects that use the Internet.

Additionally, we have instituted a new procedure for sending files between CDPHE and our hosting service located in Kansas. We now use a version of WS_FTP software (commercially produced) that encrypts files that are sent back and forth. Thus, as the hosting service sends us copies of our production files daily, the files are secure during transmission.

Please let me know if you have any other questions.

Office of Information Services, HCFA
cin:internetsecurity@hcfa.gov

The purpose of this memorandum is to file an Acknowledgement of Intent to use the Internet to transmit HCFA Privacy Act-protected and/or other sensitive HCFA information. This Intent is being filed by the

Colorado Department of Public Health and Environment
Bob O'Doherty, Chief Information Officer
CHEIS-ADM-A1
4300 Cherry Creek South
Denver, CO 80246

(303) 692-2249
Bob.Odoherty@state.co.us

The Colorado Department of Public Health and Environment (CDPHE) is a subcontractor to the Colorado Department of Health Care Policy and Financing, (Colorado Medicaid) for provision of EPSDT administrative case management services. CDPHE in turn contracts with county public health agencies in Colorado. These agencies provide assistance, and other support services that increase access to Medicaid services. These services are provided under public health nursing supervision.

CDPHE is developing new software, Integrated Registration Information System or IRIS, to be used by local public health agency staff in providing these EPSDT case management services. This software provides a listing of state EPSDT eligible clients, with Medicaid spans of eligibility, client names and

addresses and other demographic information. It also includes a "chart" area where staff can record contracts on behalf of clients, concerns, diagnoses, services and referrals. This information is used to report back to Colorado HCPF regarding activities and accomplishments under the subcontract.

Key features include a "tickler" module to allow staff to set up reminders for client activities, and a letter-writing module which uses Word to merge client data with letter templates staff can edit.

IRIS will be shared with the Health Care Program for Children with Special Needs (HCP), which has an agreement with Medicaid around certain services for special needs children. Additionally, approximately half of the HCP children are Medicaid recipients. IRIS will also be used for Women's Health programs. Initially the Title X family Planning will use the software and we hope to include the Medicaid Prenatal Plus Program as well.

The software will be loaded onto an Application Service Provider server running in a Citrix environment. Local staffs will use a Citrix client loaded on their PC to access IRIS via the Internet. IRIS includes internal security, which governs which data a particular staff member can access. Individual staff members are granted access to data according to a "need to know" basis. Additionally, training on confidentiality training and approval procedures are a part of IRIS training prior to deployment.

The identification process involves direct personal contact exchange of passwords and identities in most cases (handled during training) with some telephonic identification of users and/or passwords as necessary. All authorized users will be required to have separate user ID and passwords for access to the Citrix server. The environment will be maintained as a separate installation to isolate it from other applications.

We believe that our networking model best fits the "large organization" model in the HCFA policy. The key difference is that the firewall sits between the Citrix application server and the Internet. To protect transmission of information across the Internet IRIS will be set up to required encryption of the data stream between the Citrix client and server.

This encryption used is the Citrix Secure ICA implementation. SecureICA uses the RC5 encryption algorithm from RSA Security, Inc. The Citrix server and ICA client use the Diffie-Hellman key agreement algorithm with a 1024-bit key to generate RC5 keys.

SecureICA offers the following features:

- 128-bit encryption during user authentication
- Strong session encryption and flexible encryption support
- Per-connection encryption support

The Secure ICA server and client generate unique RC5 keys for each connection. A system service periodically generates new Diffie-Hellman parameters in the background, providing for an enhanced level of security

We feel that this implementation meets the intent of using "self-authentication, as in internal control of symmetric private keys" as specified in the HCFA Policy. However we would like to communicate with security staff at HCFA to discuss HCFA's definition of this method.

IRIS II is to be deployed beginning March 1, 2002.

APPENDIX U: COLLABORATIVE PROJECTS

Advisory Council on Health Programs for Women and Children
CDPHE Interdepartmental Immunization Initiative
Child and Infant Mortality Review Team
Colorado Cancer Registry
Colorado Covering Kids Coalition
Colorado Fetal Alcohol and Substance Abuse Coalition
Colorado Hearing Screening Advisory Committee
Colorado Newborn Screening Program Advisory Committee
Colorado Perinatal Care Council
Colorado Responds to Children with Special Needs Advisory Group
Early Intervention Task Force
Folic Acid Task Force
Healthy Child Care Colorado
Maternal and Child Health Funding Methodology Task Force
Maternal and Child Health Mortality Review Grant
Part C Interagency Coordinating Council
Prematurity Prevention Task Force
Program for Children with Special Health Care Needs Physician=s Advisory Board
Safety Net Project
Sickle Cell Advisory Group
State Medical Assistance and Services Advisory Council
Turning Point Initiative

APPENDIX V: GUIDELINES FOR CLINICAL GENETIC SERVICES

COUNCIL OF REGIONAL NETWORKS FOR GENETIC SERVICES**:** GUIDELINES FOR CLINICAL GENETIC SERVICES FOR THE PUBLIC'S HEALTH

I. ORGANIZATION AND ADMINISTRATION

A. State/Territorial Genetics Coordinator/Educator. Each state should identify a genetics unit or, at least, a full-time genetics coordinator/educator with a background in service delivery, genetics, and public health issues. The responsibilities of the coordinator should include:

1. a) Facilitating communication within the genetics community providing existing genetic services in the state/territory.
   b) Maintaining linkages between them, consumers and all relevant components of the state/territory Department of Health.

2. Familiarity with all aspects of clinical and laboratory components of genetic services including:
   a) prevention
   b) dissemination of information (training and education programs)
   c) needs and resources
   d) mechanisms of reimbursement

3. Understanding how genetic services are distributed within their state and promoting the accessibility of these services to all who need them.

4. Identifying needs for additional genetic services in their state.

5. Understanding existing data collection programs and addressing additional needs.

6. Monitoring state legislation and regulatory efforts directed at genetic issues.

7. Familiarity with recognized professional standards for clinical and laboratory personnel, facilities, and genetic services.
8. Monitoring all contracts related to state-funded genetic services.

9. Collaborating closely with the State/Territorial Genetics Advisory Council.