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The Genetic Services Plan for Wisconsin can also be viewed on the world wide web at

[www.slh.wisc.edu/genetics](http://www.slh.wisc.edu/genetics)

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## EXECUTIVE SUMMARY

*“Genetics is everywhere”* and it concerns ALL of us. Research such as the Human Genome Project is quickly bringing genetics into the forefront of medicine, promising cures for disease through gene therapy and more effective drugs that are tailored to a person's genetic make-up. Genetics is expanding beyond issues related to birth defects and rare genetic disorders that primarily affect children. We are learning that most disease is caused by an underlying genetic susceptibility that is then modified by a host of environmental factors. Genetics is becoming integrated into healthcare across the entire life cycle as more and more is learned about the role of genetics in disorders such as asthma, diabetes, heart disease, and cancer. As this integration is occurring, both medical, ethical, legal, and social challenges are arising and the needs for genetic services are increasing. How will we, as a community, be able to respond to the ever-increasing impact of genetic knowledge?

A multidisciplinary workgroup funded by a grant from the Health Services and Resources Administration (HRSA) created this Plan. The Plan is directed to all individuals who have a stake in the future of medical genetic services in Wisconsin. These include, but are not limited to, genetic professionals, primary care providers, other health care providers, governmental representatives, policy makers, legislators, educators, third party payers, and current and potential consumers.



The current system of genetic services in Wisconsin includes direct clinical/medical care services as well as activities ranging from screening programs and laboratory services, to educational activities and birth defects surveillance. Until now, planning related to how best to provide these and other genetic services has been limited. However, this type of planning is especially needed given the current explosion in genetics information and technology. The “Genetic Services Plan for Wisconsin” is one step in the process of planning for the future provision of genetic services for the people of Wisconsin.

### WHAT IS THE “GENETIC SERVICES PLAN FOR WISCONSIN”?

The Plan is a problem-oriented, needs-identification guide for addressing current and future challenges that will likely affect the provision of genetic services in Wisconsin. Recommendations are presented and are intended to be starting points for the development of future solutions. The Plan should be viewed as a “work in progress” that will be modified periodically over the coming years.

#### Mission of Genetics in Wisconsin

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The goals of genetic care are to  
optimize health,  
decrease negative effects and  
improve quality of life  
for individuals with or at risk for  
genetic conditions,  
through appropriate medical care,  
information and support.

...

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## BACKGROUND INFORMATION ABOUT GENETICS AND GENETIC SERVICES.

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- **Is genetics really “everywhere”?** Genetics can have an impact at any point in the life cycle—before and during pregnancy, in newborns, in children, in adults.
- **What does “genetic care” involve?** Two primary approaches have been involved in the provision of comprehensive genetic care: the medical model and the counseling model.
- **Who provides genetic care?** Currently, individuals with special training and expertise in genetics provide a large portion of genetic care. However, as more is learned about the role that genetic factors play in various disorders, many different specialists will likely assume new roles as interpreters of genetic information for their patients. In addition, genetic support groups and peer group organizations currently are and will continue to be integral parts of the genetic care provider network.
- **Guiding principles of genetic care provision.** Any plan for genetic services must be consistent with fundamental principles of all public health activities—family centered care, community-based access, leadership and collaboration, utilization of family resiliency, culturally competent care.
- **What are the benefits of genetic care?** Expected benefits of comprehensive genetic services include educating patients and families, assuring informed, autonomous decision-making, and providing anticipatory guidance for health care.
- **Why should genetics be in the public sector?** Sustaining involvement of the public sector in medical genetics is important for many reasons—emphasis on prevention, need for expertise unavailable in the private sector, need for public financial support, need for assurance of access to care, screening, education, policy development and surveillance.
- **Structure and services.** Currently, Wisconsin has both public sector (e.g.

University-based) and private service providers who make available several different components of care including: comprehensive clinical care, screening programs, laboratory services, educational activities, and birth defects monitoring.

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## ASSESSMENT OF CURRENT PROGRAMS IN MEDICAL GENETICS IN WISCONSIN.

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The current status of genetic activity in Wisconsin was compared with guidelines developed by the Council of Regional Networks of Genetic Services. Based upon this comparison, strengths and weaknesses of current genetic service programs were identified.

- **Strengths.** Some of the identified strengths are in the areas of: outreach programs for care; educational efforts; newborn screening; comprehensive care of “traditional” genetic disorders.
- **Perceived weaknesses and challenges.** Problems and challenges that need to be addressed in greater detail were divided into two primary areas—those that relate to current service needs and those that arise because of anticipated future challenges.
  - **Current service needs.** Problems related to current service needs were categorized into seven main issues:
    1. Need for ongoing oversight of clinical genetics activities.
    2. Inadequate documentation of needs for services.
    3. Insufficient genetic workforce.
    4. Lack of adequate funding.
    5. Challenges of providing equitable care and access to care.
    6. Need for stronger collaborations among geneticists and with other health care providers.
    7. Need for additional educational activities.
  - **Challenges for the future.** Problems and challenges related to anticipated challenges for the future were categorized into four main issues:
    1. The new genetics.
    2. Challenges of the healthcare marketplace.
    3. Transitions to adulthood.
    4. Adult-onset disorders.





**Vision for the Future**

Implementation of the principles identified in this plan will result in the seamless integration of genetic knowledge and care across the continuum of medical care delivery in Wisconsin. This will be accomplished through establishing new collaborations and alliances, providing ongoing education of all care providers, and crafting new approaches to the organization and funding of genetic services.

**EXECUTIVE SUMMARY**

**RECOMMENDATIONS**

Recommendations presented in the Plan are not intended to be specific solutions. Instead, they are to be viewed as guidelines for directions and approaches to finding solutions to the numerous issues that face genetic services in the future. Recommendations are organized into nine primary clusters:

1. **Advisory Council.** An Advisory Council for Genetic Services should be established. This Advisory Council should be charged with continuing the assessment of the status of genetics and providing guidance regarding Plan recommendations.
2. **State Presence.** There is a need to consider where genetics should be placed in the state public health organization. Consideration should also be given to establishing positions for a State consumer/patient liaison and a State genetic epidemiologist.
3. **Service Issues—Assessment and Quality Assurance.** The Advisory council should devise means of assessing current genetic services.
4. **Service Issues—Insufficient Genetics Workforce.** A survey of current genetic care provision and how it is meeting the existing needs is a necessary first step in order to address the anticipated future genetic needs of our state.
5. **Service Issues—Financing Genetic Services.** Options for additional funding must be explored. Without additional funding, access to genetic services will be limited and the ability of genetic providers to respond to the explosion of new information and new options will be severely hampered.
6. **Service Issues—Access.** Access to appropriate, comprehensive, and longitudinal care regardless of health insurance needs to be assured.
7. **Collaborations.** Steps should be taken to improve collaborations among genetic professionals as well as between genetic professionals and others (e.g. non-genetic

health care providers, managed care organizations, families, support groups, advocacy organizations).

8. **Issues Related to the New Genetics.** New genetic information and discoveries will likely change the face of genetic services, as well as fundamentally change the practice of medicine in general. For genetics, most pressing issues will be related to adequate education of primary care providers and to manpower limitations. Both of these issues will need to be addressed to adequately care for all Wisconsin families.
9. **Education Related Issues.** Educational initiatives are fundamentally important to the future viability of genetic services. Educational efforts should be directed towards physicians, other health care providers, administrators, governmental representatives, legislators, the public and those in need of direct genetic services.

We fully recognize that these recommendations are too numerous and too challenging to be addressed simultaneously. The Advisory Council will have as its first task further prioritizing these needs.



Genetic counselors often use diagrams like this one to help explain genetic concepts.

GREEN BAIN PRESS/SCIENCE

# Genetic Services Plan for Wisconsin

## INTRODUCTION

Medical genetics is a young specialty. Although its roots reach back over millennia, genetic services have been a part of clinical medicine for only the past few decades. In the past, clinical genetics professionals primarily addressed issues related to birth defects and “traditional” genetic disorders<sup>1</sup>, conditions that while individually rare, collectively affect around 3–4% of the population. Although problems related to access, service delivery, and funding existed, most problems seemed manageable given the limited scope of services provided.

Today a new face of genetics is emerging. The rapid advances in genetics due to the Human Genome Project and other scientific endeavors create challenges far greater than those faced in the past. Genetics has effects across the lifespan; genetic principles cut through every medical subspecialty; advances in molecular diagnosis and treatment will challenge health care providers and the public, ethically and morally. **“Genetics is everywhere.”** How can we respond to the challenge of providing high quality genetic services to the citizens of this State? This document is a first step in creating a plan of action for the continued development of genetic care in Wisconsin.

## WHY A STATEWIDE GENETIC PLAN?

Genetic services have always had a public health focus. Prevention (both through education and long term anticipatory medical care) and health promotion (primarily for populations with special health care needs) have been central to clinical genetics activity. Furthermore, Wisconsin has been a leader in assuring access to quality genetic services. However, planning related to such services has been limited. Therefore, a Statewide Genetic Plan is visualized as a way to assess how current needs can better be addressed and how the new challenges of the future can best be faced.

## PROCESS AND INTENDED USE

Supported by grant funds from the Maternal and Child Health Bureau, Health Resources and Services Administration, a workgroup of 34 individuals was formed (see Appendix I) in

2000 to develop a Genetic Services Plan for Wisconsin. Prior to convening the workgroup, the existing status of genetic activity in Wisconsin was compared with guidelines developed by the Council of Regional Networks for Genetic Services (see Appendix II). Through the workgroup and its subcommittees (Finance; The New Genetics;

Documentation and Data; Care Delivery; Client-Centered Care; Education and Information; State Structure), central issues were identified and solutions sought. This document is the result of these deliberations.

The workgroup determined that this Plan should be “needs identification” focused rather than “solution” oriented. Recommendations are presented, which are intended to be starting points for the development of future solutions, rather than being a fixed set of guidelines for action. The workgroup views this Plan as being a work in progress that will serve as a template for future actions and that periodically will be modified over the coming years.

## INTENDED AUDIENCES

This document is directed toward everyone who has a stake in the future of medical genetic services in Wisconsin. Included in this group are genetic professionals; primary care providers and other health care professionals; state agency staff; legislators; educators; third party payers; family and health care advocates; and current and potential “consumers.”<sup>2</sup> In addition, a primary intent of this document is to serve as a guide for the Advisory Council on Genetic Services, which the workgroup visualizes as carrying on its mission in the future.

## The Mission of genetics in Wisconsin

The recommendations generated in this report are based on this mission statement:

The goals of genetic care are to optimize health, decrease negative effects and improve quality of life for individuals with or at risk for genetic conditions, through appropriate medical care, information and support.

It is important to emphasize that **the care of the individual and family comes first.** This is an underlying principle in all recommendations.

1. “Traditional” genetic disorders refers to conditions that are rare and primarily affect children.

2. Genetic service consumers include those who need or may need genetic services and those who benefit or may benefit from genetic education activities.



## BACKGROUND INFORMATION ABOUT GENETICS AND GENETIC SERVICES

### AN INTRODUCTION TO GENETICS

**“Genetics is everywhere.”** Genetics can mean many things. In medicine, genetics can refer to changes in genes; it can refer to disorders that are passed on in families; it can refer to birth defects and their causes; and it can refer to complex conditions where genetic and non-genetic factors play a role in the development of a disorder, such as diabetes, heart disease and cancer. Medical genetics deals with all of these issues and their impact on individuals and families.

Genetics can have an influence at any point in the life cycle:

#### Before pregnancy and during pregnancy:

- Neural tube defects, such as spina bifida and anencephaly, are birth defects that are caused by a combination of genetic and environmental factors. Adequate intake of the B vitamin folic acid has been shown to prevent up to 60–70% of all neural tube defects. Genetics professionals are involved in educating the public about the importance of taking folic acid before and during pregnancy.
- At least 20% of all recognized pregnancies end in miscarriage. Genetic factors (mainly chromosome abnormalities) cause at least half of all of miscarriages. Understanding the genetic cause of miscarriage can often help couples cope with their loss and provide important information on implications for future pregnancies.
- Exposures during pregnancy, such as to high levels of alcohol, can affect a baby's development and result in birth defects that could have been prevented. Teratogen Information Services, which are often affiliated with clinical genetic centers, provide the general public as well as health care professionals with information on pregnancy exposures and birth defects prevention.
- Prenatal screening, such as use of maternal serum triple screening, can identify pregnancies at risk for disorders such as neural tube defects and Down syndrome. Genetic counseling can educate patients about potential risks to their pregnancies and allow them to make informed decisions about prenatal diagnostic testing.

- Prenatal diagnostic tests, such as amniocentesis and ultrasound assessment, can provide families with information about the risk (or lack of risk) that their baby may be born with certain birth defects.

#### In newborns:

- At least 3% of all babies are born with physical birth defects, most of which are genetically determined. Physician geneticists are involved in determining if a birth defect is isolated or associated with other medical problems. This knowledge can be important for the care of the infant and may have medical implications for other family members.
- Newborn screening tests are a powerful tool for early identification and treatment of certain disorders. However, such screening and treatment methods are currently available for only a few of the causes of birth defects and genetic disorders.

#### In children:

- Learning disabilities and mental retardation affect at least 5–10% of the population. Most individuals who are mentally retarded have genetic causes for their disability. Identifying a cause is essential for accurate genetic counseling of other family members regarding potential risks for having a child with similar problems.
- Genetics also plays a role in the development of many common medical disorders of childhood, such as asthma and juvenile diabetes. Understanding the genetic causes of such common childhood disorders is helping to develop better prevention and treatment strategies.

#### In adults:

- **In the past, medical genetics focused on disorders of infants and children. More recently there has been an explosion of information about genetic involvement in common adult-onset conditions.** Breast and ovarian cancer, colon cancer, heart disease, adult-onset diabetes, mental illness, hemochromatosis and a host of other disorders are now recognized as being caused, at least in part, by genetic factors.



Justin and his parents meet with Dr. Pauli (see Lifeline at right).

# LIFE l i n e



## The Importance of a Diagnosis

Our family's story—Justin's story—began when Justin was just four months old. He became upset and hard to comfort, sleeping only if I held him in my arms. When our pediatrician saw him the next morning, examination and testing didn't show any reason for his fussiness. Although the pediatrician suspected an infection, two more exams over the next two days failed to really uncover what might be going on.

On the fifth day of his apparent illness, I noticed that Justin's leg seemed hard and swollen and that he wouldn't move it. Our pediatrician scheduled a CT scan, thinking that an infection might have moved into Justin's leg bone. However, when we went to have this done, the radiologist looked at Justin's leg, ordered an x ray and then informed us that Justin had a broken leg. Our nightmare had begun.

They took Justin to the ER. My husband, Bill, and I were upset and crying. An orthopedist asked us a lot of questions and asked that a skeletal survey be done. It showed that Justin had broken ribs, too. We knew then that there was something medically wrong with our baby, but we didn't know what it was. And, everyone else seemed to think that he had been injured on purpose. Justin was taken to the operating room and we went to the waiting room...to wait. A police deputy arrived and asked us a series of questions. A social worker questioned Bill and me separately. The deputy questioned us again! The same questions, the same answers, but it was as if our answers weren't satisfactory.

The next day, Bill and I were total wrecks. We were both crying. We were very anxious to see our own pediatrician. After all, he knew us, had taken care of our older son; he would make them understand that we really didn't know how Justin got those broken bones. He didn't say much at first. Then he told us that he would have to agree with the surgeon that the injuries were not accidental! I asked him if Justin could have a disease that caused this, and he said no. Bill and I were hysterical now. Bill asked our pediatrician what was going to happen now, "are they going to take my kids away?". The pediatrician just said he didn't know what was going to happen. It dawned on us that we were going to need legal help.

Later that day another social worker came by. She tried to act like she was our friend. She said that she had no kids but knew having two children can be stressful. She kept repeating that she was there to help us. Then she told us that she knew what we were going through. She had no idea! She had us sign some papers that said that a grandparent had to be present when Bill was home with our older son, Jason, or they were going to take him away and put him in a foster home. When she left we realized that we might lose our boys. What was to happen to our family? I was so upset and scared. All I could do was cry and hold Justin.

We arranged for protective custody in our home with grandparent supervision at all times. We had to agree. It was nearly time for Justin to be discharged and he had to be able to come home with us.

The detective came to visit again. He told us that things did not look good for us. He said we have a baby with multiple fractures and no explanation and that the doctors say that this is not a bone disease. He looked right into my eyes and stated that he knew one of us did this and that he thought he knew which one it was. He said he was willing to go to the DA and state that this person did this but now had remorse

if one of us would admit that we had injured Justin. He told us that he had warrants for our arrest. I couldn't believe what I was hearing. I was so scared. Arrested for something we didn't do? Worse, all of these people thought we would purposely harm our baby. It was like a bad nightmare. When were we going to wake up?

When Justin was finally discharged, we all got to go home, but everything wasn't just fine. In fact, nothing was fine. Our attorney had told Bill that someone might be doing jail time. Jason seemed frightened of us. I felt like I was falling apart. I remember that Bill got me into the shower and when he came to check on me, I was just sitting in the tub with the shower running over me. He helped me out. He took me to see our Pastor. I don't remember much of the meeting with Pastor, but at some point that night I knew we had to fight to keep our family together. Somehow we had to find the help that our baby needed.

Bill's sister had found information on the Internet about osteogenesis imperfecta. I could not believe what I was reading. Justin seemed to have all the symptoms and signs. Even though the doctors had told us Justin didn't have a bone disease, we needed to get him to a specialist. I contacted a support group and through them got the name of such a specialist—Dr. Pauli, a geneticist in Madison. I called and talked with him, but it would be almost two weeks before he could see Justin. We lived in chaos for that time. Supervision at all times—what does that mean? Bill couldn't even take Jason to Sunday School without a chaperone. When I nursed Justin, someone had to be there with us. Jason changed from his usual happy-go-lucky 3 year old and had his own times of crying.

When we went to the Clinical Genetics Center at the University of Wisconsin, Dr. Pauli wanted to hear our story. Then he examined Justin as best he could—mostly from the waist up since Justin was still in a spica cast. After that he told us that he was almost certain that Justin had osteogenesis imperfecta, probably type I. Both Bill and I began to cry again—partly from relief that we finally knew what was the matter with our baby and partly in fear of what that meant for our beautiful son. Dr. Pauli talked with us about safe handling of a baby like Justin. He wrote a letter for us to take home with us and told us that he hoped that it would help in our dealings with the social workers and the police. I honestly thought that everything would be dropped the next day. Little did I know! In fact, it was six more weeks before the abuse charges were fully dismissed.

A skin biopsy confirmed that Justin has type I osteogenesis imperfecta. Now we have to face the future of caring for a son with a disorder that places him at risk. It isn't a happy thing to be told that your son has a problem that will affect him for the rest of his life, but we know that we'll have the medical help we will need to care for him. Without the services of a geneticist like Dr. Pauli and the others at the Clinical Genetics Center who knows how this part of Justin's story might have ended.



- **Notably, a major effect of the “new genetics” is recognition of the complex genetic factors that can predispose each of us to a specific set of adult-onset disorders.** Individuals who are aware of their increased genetic risk for certain conditions can work with health care professionals to schedule appropriate intervention and surveillance strategies, which allows for early identification and possibly prevention of the disorder.

Indeed, “*genetics is everywhere.*” The needs for genetic services will increase as the Human Genome Project matures and we are able to incorporate this new molecular information into medical care. As a community, how will we be able to respond to the ever-increasing impact of genetic knowledge?

### COMPONENTS OF GENETIC CARE

Traditionally, the provision of genetic care has involved two primary approaches: the “*medical model*” and the “*counseling model*”. The medical model focuses primarily on clinical diagnosis, medical prognosis, and ongoing health care needs. The counseling model focuses on providing information, choices and support in a “non-directive” context. Both approaches rely upon appropriate identification and assessment of the genetic issues involved.

The second approach, called genetic counseling, is less common in routine clinical practice and often misunderstood. Genetic counseling is a specialized communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. As members of a health care team, genetic counselors provide information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. Genetic counselors identify families at risk, investigate the problem present in the family,

interpret information about the disorder, analyze inheritance patterns and risks of recurrence, and review available options with the family. Genetic counselors also provide supportive counseling to families, serve as patient advocates and refer individuals and families to community or state support services. Genetic counselors serve as educators and resource people for other health care professionals and for the general public.<sup>3</sup>

Though the medical model and counseling model each have independent value, the needs of individuals and families affected by genetic conditions are best served when these two models are combined in the provision of comprehensive care (see diagram below).

Other components of genetic service reach beyond patient- and family-based clinical care, including:

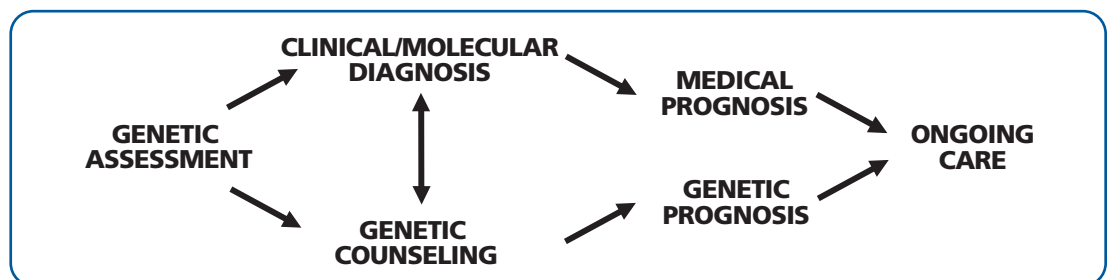
- Laboratory support
- Screening of populations at risk
- Teaching and training
- Public education.

Public education is particularly crucial in providing the public with the resources to address genetic issues relevant to their lives.

### PROVIDERS OF GENETIC CARE

Genetic professionals provide health care services to families and offer other services, such as laboratory support.

- **Clinical Geneticists** are physicians with training in a primary specialty (e.g. pediatrics, obstetrics, internal medicine) and additional subspecialty training in the clinical aspects of genetic care. Medical geneticists receive board certification through the American Board of Medical Genetics.
- That board also certifies the training and expertise of **Clinical Biochemical Geneticists** (specialists in the diagnosis and treatment of inborn errors of metabolism), **Clinical Molecular Geneticists** (experts in the use of molecular tools in



3. Adapted from National Society of Genetic Counselors website, [www.nsgc.org](http://www.nsgc.org)



## The Role of the Genetic Counselor

I am the proud wife and mother of two—Kymberley, 6 years and Mikey, 22 months old. Kym has been a healthy child, overall; Mikey has not. When Mikey was 10 months old, I started to notice that his development wasn't where it should be. I can't count the different doctors, pediatricians and specialists that we had been to. I did finally find a doctor who was willing to listen to my concerns with Mikey. He, too, had noticed that there were delays. I then got involved with the Birth to Three Program, which has been wonderful. That is how we got Mikey started with therapy (he couldn't sit up or hold his bottle by himself.)

Because of his eating habits, the doctor had referred us to a speech pathologist who evaluated Mikey. After the evaluation she talked with our doctor and confirmed his suspicion. I knew my son was delayed, but never in a million years was I ready for what the doctor was about to tell me. The doctor said "I think Mikey may have Cerebral Palsy." I cried. We then got referred to a neurologist.

On our first visit to neurology another bomb was dropped. He said that Mikey didn't have Cerebral Palsy. We were back to square one again, not knowing what was wrong. The neurologist suspected Autism. He had given us some information, but didn't want to give us too much information until we knew for sure. I cried again out of frustration for Mikey. Our second visit consisted of all the testing (I can't remember them all). This was hard because we still knew nothing and I was scared, not only for me, but more for Mikey.

It had been a few weeks and we were waiting for the test results. The phone rang—it was a genetic counselor. She had results of the tests. We learned that Mikey has Fragile X syndrome. I can remember feeling lost and relieved all at once. I was in shock. I cried. The counselor explained what Fragile X was and its cause. She sent me some information and gave me some websites to check out. She made me

and my son feel important.

We then had a consultation with the genetic counselor, Theresa, and she explained Fragile X in "English." She gave us so much information and answered all of our questions. She was great! She included my daughter in the conversations and suggested that eventually we might want to get her tested, too. We have to remember that Kym is affected by Mikey's diagnosis, and Theresa did.

We set up an appointment with a geneticist, Dr. Wargowski and his staff. I was impressed and overwhelmed by how helpful and observant everyone was. I don't think there was anything that they didn't check. They had also included Kym and examined her, too. We had made the decision that we would get her tested. This is when I finally got to meet Sumedha, the second genetic counselor. I had talked to Sumedha many times on the phone and she made me feel like I was the best mom in the world. She encouraged me, listened to me and guided me in several situations. The first thing she did was give me a big hug. She knew that I was overwhelmed by everything going on in the last month.

I was nervous about Kym and her results. I was scared that I was going to get that call again. We got the call from Sumedha and to my surprise it was negative! Sumedha was almost as excited as I was. We actually got to share a moment of joy.

I can't put into words what the whole "genetic department" has done for my family. My family went through a lot of emotions, and they were the ones that supported us. I appreciate them and their hard work. Thanks to Sumedha and Theresa I am learning all I need to know about Fragile X and my son.



the diagnosis of genetic processes), **Clinical Cytogeneticists** (experts in the utilization and interpretation of chromosome analyses) and **Ph.D. Geneticists**.

- **Genetic Counselors** have masters-level degrees and are trained in all aspects of medical and clinical genetics as well as in counseling. Counselors are certified through the American Board of Genetic Counseling.

As yet there is no mechanism for certification of clinics and sites that provide genetic care. However, training sites for medical genetics specialties and genetic laboratories must be certified.

In addition to these care providers, various aspects of genetic services may be made available by other health care providers such as perinatal specialists involved with prenatal diagnostic services and nurses with advanced

training in genetics. As the understanding of genetic predispositions to various disorders is better worked out, many different specialists will need to assume new roles as interpreters of genetic information for their patients.

Genetic support groups and peer group organizations can provide another kind of care—care extending beyond the clinical setting. **Only if families and support groups are recognized as an integral part of the care provider network can genetic care be truly comprehensive and therapeutic gains maximized.**



Mikey and his Mom meet with genetic counselor Sumedha Ghate.



## GUIDING PRINCIPLES IN GENETIC SERVICE PROVISION

Any plan for genetic services should be consistent with fundamental principles of all public health activities.<sup>4</sup> These include:

- **Family Centered Care.** Caring for the entire family has been central to genetic services since their inception. Genetics, after all, fundamentally deals with issues that may impact many current and future members of families. Furthermore, the founding principle of providing information in a non-directive context emphasizes empowerment, respect and autonomy.
- **Community-Based Access.** Outreach activities have been and remain a central part of genetic care in Wisconsin. Such outreach not only provides services near the communities in which families live, but also strives to assure equality of access to services and to function as a “safety net” for families who otherwise would be without this help. Additionally, geneticists often must assume a major role in the coordination of care of complex conditions. As the role of genetics in medical care expands, the challenges of providing equitable access to this care will also increase.
- **Leadership and Collaboration.** As reflected in the development of this document, the genetics “community” must include not only care providers but all others who have a stake in assuring that quality services are available to all people of Wisconsin—including families, community leaders, and governmental representatives.
- **Utilization of Family Resiliency.** Here, too, the tradition of fostering autonomy and empowerment places genetic care provision at the forefront of such a guiding principle.
- **Culturally Competent Care.** Genetic services in Wisconsin have customarily been provided in a setting that values diversity and respects individual beliefs and family traditions.

## BENEFITS OF GENETIC CARE

Why should genetic care be considered a crucial partner in medical care? What benefits result from such care? The following is an

outline of the expected benefits of comprehensive genetic services:

- **Family and Patient Education.** Often family members do not fully understand a diagnosis and its implications until they meet with a genetics professional.
- **Informed Decision-Making.** Autonomous decision-making—about health care as well as reproductive decisions—can only be made when the implications of those decisions are fully understood. The non-directive manner of genetic care seeks to assure that the decisions families make will be fully informed and, ultimately, their own.
- **Anticipatory Guidance.** Specific diagnosis is the cornerstone of preventive care in genetic disorders. Once a diagnosis is established, whether by testing or clinical assessment, the affected individual or family can be educated regarding the prevention of future medical problems and the identification of risk in other family members. Presymptomatic genetic diagnosis and predictive testing for predisposition to disorders such as cancer and heart disease can provide an opportunity for anticipatory guidance and risk reduction; this not only will help the affected individual and family, but also can reduce overall health care costs.

## WHY SHOULD GENETICS BE IN THE PUBLIC SECTOR?

In contrast to most other medical subspecialties, medical genetics has traditionally straddled the public and private domains of medicine. In part this was simply of necessity, since reimbursement for many genetic services is inadequate to assure their continuing existence without public funding. In addition, genetic programs are in many ways similar to other public health programs.

Following are some justifications for sustaining involvement of the public sector in medical genetics:

- **Emphasis on prevention.** Medical genetics emphasizes preventive aspects of care that are also central to public health initiatives. Preventive services in genetics can be divided into primary, secondary and tertiary levels.
  - Initiatives to ensure adequate *folic acid supplementation* as a means of prevent-

## Ongoing Genetic Care

Our son Esteban was born in 1980. At first, he seemed like any other “normal” baby. He crawled in an army type fashion, lying down and using his arms to drag himself. He walked by 11½ months. He was speaking clearly at an early age. It wasn’t until he was about 1½ years old that we started to notice some “problems”. First, the baby fat on his hands was not going away. Then, his hands began to show signs of stiffness. After this, he started to struggle with his fine motor skills - he couldn’t pick up Cheerios as easily as before. Holding on to cups and glasses became a problem – the cups and glasses would, many times, slip from his hands and fall to floor. Esteban also began to have frequent ear infections that eventually caused hearing problems - thank God his speech was well developed before this occurred and he was able to communicate clearly in Spanish. Because of these struggles, we sought the advice of our pediatrician who recommended that Esteban be seen at a genetics specialty clinic located in Children’s Hospital of Wisconsin.

Here we met Ms. Lu Ann Weik, a genetics counselor. She became and still is our link to many of the services and opportunities that Esteban has been fortunate to receive. At our initial visit, it was suspected that Esteban might have some form of arthritis. Esteban was then seen by Dr. Herrmann, a geneticist, who first expressed to us that Esteban might have a rare genetic condition. After many tests and a visit with another geneticist, we were finally told that Esteban does have a rare genetic condition called Hurler-Scheie Syndrome. As I sat in the doctor’s office with Esteban in my arms and was being told of his condition and the expected prognosis, I felt devastated and crushed. My worst fears were confirmed as the doctor explained that Esteban most likely would not be able to do any physical work and possibly could be in a wheelchair by the 8th grade. He continued by saying that there was nothing that could be done to stop the progression of the illness. My child’s health, I believed at the time, was about to deteriorate quickly. I also believed that there was nothing I would be able to do to stop or even improve what was expected to happen. I felt absolutely alone and numb with pain. Even after all these years it is difficult to relive.

However, we were and continue to be fortunate to be connected with the Genetics Department at Children’s Hospital of Wisconsin. Because of people like Ms. Weik and Dr. Lubinsky (Esteban’s pediatric geneticist for several years), Esteban and our family have not been alone and we have been able to fight this condition. Early on, Ms. Weik took it upon herself to serve as an educator, an advocate, and a friend for Esteban and our family. She helped us understand what Hurler-Scheie is, she connected us with helpful resources, she answered our questions, and she has continually provided support and encouragement. We also met annually with Ms. Weik, Dr. Lubinsky, and a team of other health care providers (including other specialists and physical and occupational therapists) to plan out a strategy for Esteban’s health; this continued without much persistence from my part from the time Esteban was diagnosed at the age of four till he turned 18. More recently, Ms. Weik and the Genetics Department are assisting us while Esteban is involved with a research study that presently requires us to travel weekly to New York so Esteban can receive special treatments.

If not for the ongoing, consistent efforts and advocacy of people like Ms. Weik, Children’s Hospital of Wisconsin’s Genetics Department, and Dr. Waters (Esteban’s pediatrician), Esteban would not be where he is today. Their assistance, guidance, support and encouragement over the past 20 years have helped improve Esteban’s health and quality of life. They have helped Esteban and our family through at least 14 different operations and procedures to help correct different problems such as preventing stiffness and improving flexibility in his hands, reducing pain in his wrists, and reducing occurrences of chronic otitis media. They also have given us hope and encouragement for the future. In spite of his “condition” and the current weekly trips to New York, Esteban is active and continuing his education at the University of Wisconsin-Milwaukee. We feel confident that the assistance, guidance and advocacy already put forth from the Genetics Department at Children’s will continue and that Esteban and our family will have access to this wealth of resources for many more years to come.

ing many instances of neural tube defects are an example of *primary prevention*.

- *Secondary prevention* includes *newborn screening* for selected congenital disorders, which, while not preventing the disorder itself, can eliminate or modify the effects of these disorders.
- Often *tertiary prevention* is overlooked—comprehensive expert *care coordination* uses evaluation and intervention in order to decrease or prevent the negative effects of a disorder.

• **Need for expertise unavailable in the private sector.** While collectively common, many genetic disorders are individually rare. Comprehensive, expert care of such disorders

is often unavailable except through public support of specialized programs.

• **Need for public financial support.** It has been well documented that certain genetic services are grossly under-reimbursed.<sup>5</sup> Compared to other medical services, genetic services are very time-intensive and reimbursement is most often about ¼ of the actual cost of their provision. Furthermore, no method currently exists in Wisconsin to secure reimbursement for most of the services provided to patients by genetic counselors.

• **Need for assurance of access to care.** Certain aspects of genetic care (such as prenatal diagnostic testing) may receive adequate reimbursement. Because of this better reim-

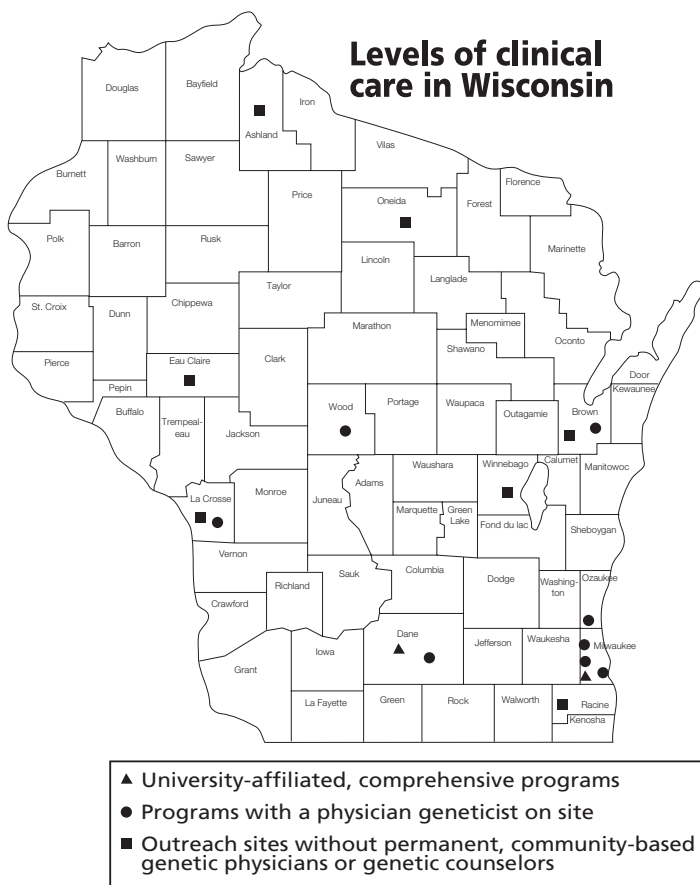
5. Bernhardt BA, Pyeritz RE: The economics of clinical genetics services. III. Cognitive genetics services are not self-supporting. Am J Hum Genet 44:288-293, 1989.



## History of Clinical Genetics in Wisconsin

There is a long, distinguished tradition of genetic services in Wisconsin, in dysmorphology (David W. Smith, John Opitz), biochemical genetics (Harry Waisman), cytogenetics (Klaus and Eva Patau) and so forth. True service delivery in genetics began in the mid-1970s and, over the course of the last 30 years, has evolved into a complex web of direct clinical care, outreach care, education, training and systems development. From those beginnings, the Wisconsin Idea—the commitment of the University to provide service to every boundary of the State—has been central to the activities of University-based clinical genetics in this State.

Block grant mandates, which began in 1983, made the State of Wisconsin, through administration of Maternal and Child Health funds, one of the major funding sources for providing genetic services. Additional collaborations have developed between University of Wisconsin based providers, their associated outreach clinics (e.g. Eau Claire, Green Bay, Neenah, Rhinelander, etc.), providers at the Medical College of Wisconsin and the Children's Hospital of Wisconsin, as well as private-sector providers (e.g. Dean Medical Center in Madison, Gundersen Lutheran Medical Center) making the public health commitment to genetics truly statewide.



bursement, private providers have been willing to offer these services. However, such private provider models have little incentive to assure access to other genetic services that are often more complex and time-intensive. Also, such models do not guarantee equal access to care regardless of economic or geographic factors.

- **Screening.** Population based screening (e.g. newborn screening, carrier detection) is a component of genetic service provision that does not easily fit most private models of care.

- **Education.** Public education is an essential part of the broader sense of genetic services, but it, too, is a component of genetic services that may not easily fit a private model of care.

- **Policy development.** Dramatic expansion in genetic knowledge and its applications require policy development as a central element of genetic service provision. Availability of new genetic technologies has raised ethical, legal and cultural issues to an extent unprecedented in recent medical history. Policy development fits best into a public health model of service.

- **Surveillance.** Birth defects monitoring and similar activities necessarily are within the public rather than private sector. These are activities that require participation of a wide range of providers from throughout the State in order to be useful.

## CURRENT STRUCTURE AND SERVICES

Over the last four decades a complex, multi-faceted set of genetics programs has arisen in Wisconsin. These include direct clinical care

8. One approach to defining levels of care is as follows:

- University affiliated comprehensive programs. Two such programs exist in Wisconsin, at the University of Wisconsin–Madison and through the Medical College of Wisconsin and Children's Hospital of Wisconsin in Milwaukee. Such programs not only provide general genetic services, but also a variety of subspecialty clinical activities (e.g. skeletal dysplasias, neurofibromatosis, sensory deficits, sickle cell disease etc.). In addition, these units are central in continuing medical education and training of professionals in genetics.
- Programs with a physician geneticist on site, providing regionally based general, and in some cases subspecialty, genetic services. Examples include the La Crosse Regional Genetics Program, and the Marshfield Clinic Genetics Program.
- Self-contained health care system based programs with a physician geneticist on site. It is likely that this model will develop in this state in the future.
- Programs with genetic counselor(s) on site that work with physician geneticists who are based elsewhere (either from level 1 or 2 programs or private providers). Examples are the programs in Green Bay and Neenah.
- Outreach sites with or without a site-based coordinator, but without permanent, community-based genetic physicians or genetic counselors. Such outreach activities are provided in Eau Claire, Ashland, Rhinelander and Racine.
- Private Providers – Various services are offered by other, private clinical geneticists and by privately employed genetic counselors. Most of these services are located in Milwaukee and the surrounding suburbs.

services, as well as activities ranging from screening programs and laboratory services, to educational activities and birth defects surveillance. Historically, service provision in genetics was primarily through University-based centers. Currently, Wisconsin has both public sector (e.g. University-based) and private service providers. Within the State, there are presently 17 board certified clinical geneticists<sup>6</sup> and 29 board certified genetic counselors.<sup>7</sup>

These providers offer different components of care including:

- **Comprehensive Clinical Care.** Various 'levels' of clinical care (reflecting the comprehensiveness of the services offered) are provided within different settings throughout the state.<sup>8</sup> (See diagram at left.)
- **Screening programs.** Wisconsin has several

screening programs, including the highly successful State directed newborn screening program, and various population-screening activities (such as for Tay-Sachs carrier identification in selected populations). With the continued progress of the Human Genome Project and related research, it is inevitable that such screening programs will expand in the future. It is likely that the Newborn Screening Program will continue to add other disorders just as hemoglobinopathies, congenital adrenal hyperplasia, and abnormalities of fatty acid metabolism have been added in the past.

Also, entirely new populations may be offered screening, such as cystic fibrosis carrier detection for all pregnant women.

- **Special programs.** Although not strictly within the confines of genetic disorders, cer-

# LIFE l i n e

## Genetic Care in the Community

We are currently parents of three children—Molly (age 7), Morgan (age 5), and Brady (age 1). Our middle child Morgan was born 5 & 1/2 weeks premature due to a very complicated pregnancy. Initially doctors thought she was a healthy 6 pounds 2 ounces baby girl but that all changed.

At just six days of age, Morgan was hospitalized for a urinary tract infection and jaundice. After that, things just slowly got worse. Each month, during Morgan's first year of life, she was hospitalized six to seven days at a time for almost anything you could think of—RSV, rota-virus, dehydration, high fevers, vomiting, respiratory distress, etc. Besides all of these complications, Morgan did not want to eat. We told our doctors many, many times that we had to force Morgan to take every ounce of milk. We felt we were starving our child, but not at all purposefully.

When Morgan was approximately eight months of age, we were referred to Madison for genetic counseling. Fortunately, the University of Wisconsin Clinical Genetics Center provides outreach services. Instead of traveling four to five hours from Holcombe to Madison, we were able to travel one hour to Eau Claire for Morgan's genetics appointment. Dr. Wargowski, our geneticist, and Wynne Cook, the project director of We're For U, the Western Regional Center for Children with Special Health Care Needs, were wonderful. After already having seen so many different professionals, we felt that Dr. Wargowski and Wynne were one of the first ones to really understand what we were going through. They seemed to believe us when we told them that Morgan plainly refused to eat. They really wanted to help us find some answers to help our child. Unfortunately, at this point, no definite answers were available (like she has "this syndrome" and "we are going to do this"). Yet, Dr. Wargowski and Wynne supported us; they made us feel like they were behind us. It wasn't time to give up hope yet.

Over the next five months, Morgan's feeding issues did

not improve. She continued to fall off the growth charts and become more and more frail. We spent more hours on the road and sought out opinions from many other professionals in the area. Finally, a team of professionals suggested that Morgan may have Noonan's Syndrome.

We thought now would be a good time to see Dr. Wargowski again. With Wynne's help, an appointment was scheduled to see him in Eau Claire. Dr. Wargowski confirmed the diagnosis of Noonan Syndrome. While many people may panic and dread hearing the word "syndrome", for us, after thirteen months of the "unknown", it was music to our ears. Wynne and Dr. Wargowski were again very supportive. They helped us learn about Noonan's. They gave our pediatrician information about Noonan's. They helped link us to support groups and other families dealing with Noonan's. A few years later, they were available when we were trying to make the decision about whether we should have another child. (We, in fact, did have another child after Morgan—a healthy and strong baby boy named Brady.)

Genetic counseling is a wonderful service; it is much more than just telling you that your child has a syndrome and convincing you from having more children. People should never be afraid to seek the expertise of genetic professionals. Morgan is now five and thriving well, but we still see Wynne and Dr. Wargowski and benefit from their knowledge. They always are available and seem more than willing to try to answer any questions that may arise. (Just last month we went back for their opinion on the growth hormone.) It is a great advantage to have We're For U and the Genetics Clinic to turn to for help and support.



6. These include 8 in the Milwaukee area, 5 in Madison, 2 in Marshfield, 1 in La Crosse, and 1 in Green Bay.

7. Currently these include 6 in Milwaukee, 18 in Madison, 3 in La Crosse and 2 in Green Bay.



tain other care programs have by tradition (and default) been directed by geneticists in this State. They include, for example, the Teratogen Information Service (for counseling of families concerned about exposures in pregnancy) and the Stillbirth Service Program (a statewide service program for evaluation of the causes of intrauterine death).

- **Laboratory services.** Most laboratory services are available nationally. However, the presence of state-based cytogenetic, biochemical genetic and molecular genetic laboratories is beneficial because of the on-site expertise that laboratory staff can offer local genetic care providers.
- **Educational activities.** Genetic professionals in the State provide hundreds of programs each year to various groups with varied educational needs.
- **Birth defects monitoring.** The Wisconsin Birth Defects Prevention and Surveillance Program is charged with developing a surveillance system to monitor the occurrence of birth defects, which will facilitate the identification of their causes and guide prevention programs. Genetic professionals within the State are integrally involved in the development and implementation of this program.

## ASSESSMENT OF CURRENT PROGRAMS IN MEDICAL GENETICS IN WISCONSIN

An essential component of the generation of this Plan was self-assessment of the current status of genetic services in public health in Wisconsin. As a first step, the Council of Regional Networks for Genetics guidelines were compared with current structure and services in this State. A detailed summary of that comparison can be found in Appendix II.

### STRENGTHS

Based on that comparison the following *strengths* were identifiable:

- **Presence of a genetic professional within the State system.** A genetic counselor currently serves as Wisconsin’s State Genetic Coordinator. This position is located in the Division of Public Health, Bureau of Family and Community Health.
- **Existence of a well-established, well-**

**respected and effective newborn screening program.** It is estimated that 99% of all Wisconsin newborns receive newborn screening.

- **Existence of active and highly successful outreach programs for care.** The Statewide Genetic Services Network provides outreach genetic services in LaCrosse, Eau Claire, Green Bay, Neenah, Racine, Rhinelander, and Ashland.
- **Emphasis on educational efforts in a variety of formats and for a variety of audiences.** These efforts include a post-doctoral training program; a genetic counselor training program; extensive CME activity; teacher-educator programs; and programs for schools, for the public, and for various support organizations.
- **Emphasis on ongoing management and comprehensive care of conditions ‘traditionally’ viewed as genetic.** Comprehensive care and specialty clinics are available for conditions such as sickle cell anemia and associated disorders, dwarfing disorders, neurofibromatosis, cystic fibrosis, genetic vision and hearing impairment.
- **Presence of special expertise regarding the care of individuals with certain specific groups of genetic disorders.** For example, biochemical genetics clinics are located in Madison and Milwaukee; craniofacial disorders clinics can be found in Madison, Milwaukee, and La Crosse; cancer risk counseling is available in Madison, Milwaukee, La Crosse, Marshfield, and Green Bay.

**Weaknesses** were also identified. Planning for the future requires identification and exploration of current weaknesses that need to be addressed in greater detail. Weaknesses can be divided into two primary areas – those that relate to *current service needs* and those that arise because of anticipated *future challenges*.

### PERCEIVED WEAKNESSES AND CHALLENGES I: CURRENT SERVICE NEEDS

**Issue 1: Need for Ongoing Oversight of Clinical Genetics Activities.** To this point, Wisconsin has had no formal oversight and planning process regarding genetic services. There is a need for an Advisory Council for Genetic Services that will engage all stakeholders in assessment, recommending change and advocacy.

## Prenatal Assessment

My husband Tony and I had decided that once we left the Chicago area and relocated it would be time to have children. In the summer of 1995 we moved to Madison and by Christmas we had a wonderful secret that we couldn't wait to share with our family and friends.

I knew a lot about pregnancy—about what could go wrong, about birth defects. This knowledge drove me to be more than conscientious and extra cautious during this pregnancy. I faithfully took my prenatal vitamins prior to and during the pregnancy. I ate healthy foods. I got moderate exercise. I avoided cigarette smoke and anything potentially toxic. During my 16th week of pregnancy, I went to the doctor for a routine blood test called the “triple screen”. I took the test and really didn't have worries that I would get bad news.

About one week later I received a call from my OB. My heart skipped a beat. I knew there was no good reason for a personal call. She informed me that one of the levels from the triple screen was elevated and asked that my husband and I come in for an ultrasound as soon as we could. I was devastated. Tony and I read our baby book. We read our triple screen handout. Our dates weren't off. I was sure I wasn't having multiples. We felt there was no glitch, no mistake; this was bad—very bad.

An ultrasound was scheduled for the very next day. It was a Thursday. I can recall the smell of the office and feel the chill I experienced lying there with a sheet draped over my belly. The technician was silent while moving the wand over my belly. Our doctor said, “I have bad news guys”. The tears were streaming down both my husbands' and my face. Our child had “anencephaly”. This was a neural tube disorder and, in our case, our baby's brain had not developed. Our child would not live after birth. I expected bad news at this visit, but nothing like this. I was numb.

Tony and I knew little about anencephaly. We didn't know that



genetics is involved.

Shortly after we lost our baby, we met with Connie, our genetics counselor. The visit answered so many of our questions, such as will this happen again, what are the odds this will happen again, what are the reasons for this happening, and what can we do to better our chances. Connie gave us an in-depth explanation about anencephaly and other neural tube disorders. She also explained to us that there was something we could do to help decrease our chances of having another baby born with anencephaly or other neural tube defect. There were no guarantees; it was all about odds and the odds were in our favor, especially if I took a simple vitamin supplement called folic acid. I was so relieved to hear that there was something proactive I could do. We left the visit with the answers we needed and with high hopes for acquiring the family we desperately wanted.

About six months later, I became pregnant again. Tony and I were happy, but the fear of finding out that this baby might have a neural tube disorder did remain in the back of our minds. We “passed” the blood test for the neural tube disorders this time around. However, one of the blood levels was low. We met with our doctor who explained that the baby could possibly have Down syndrome. We decided to have an amniocentesis to find out if there was anything wrong. We couldn't believe we were in this nightmare again. When the results came back, our fears were swept away. We had a healthy baby in utero!

Our son Joseph was born without complication in 1997. He was the greatest gift Tony and I have ever received. All of our worries and all of the bumpy times we endured with the pregnancies did not deter us from having the family we so wanted. When Joey was just 14-months-old Tony and I conceived again. We, once again, had a difficult pregnancy, but this time there were no scares of a disability. Our healthy identical twin daughters were born in 1999. What a family we now have!

**Issue 2: Inadequate Documentation of Needs for Services.** Data are inadequate to accurately estimate current needs, let alone needs for the future. Furthermore, there is no system in place to collect accurate data on individuals and families who receive genetic services. This is particularly difficult since it has proven challenging to coordinate data collection from all genetic care providers. In addition, no guidelines for minimal levels of genetic services for the State have been generated. On these bases there are currently no accurate data about what services should be provided, how extensive needs for those services are, or which service needs are currently inadequately addressed.

**Issue 3: Insufficient Genetics Workforce.** Recently Kaiser Permanente (a major health care organization serving various U.S.

regions) internally estimated staffing needs for currently appropriate genetic services.<sup>9</sup> Extrapolating those data to Wisconsin's population suggests that adequate service would require the presence of 20 clinical geneticists and 80 genetic counselors. Based on a census of currently board certified personnel in the State, currently there are approximately 12 full-time equivalent (FTE) clinical geneticists and 22 FTE genetic counselors in Wisconsin. In order to provide appropriate levels of service (not taking into consideration the issues addressed in the next section), the number of physician geneticists would need to be nearly doubled and the number of genetic counselors almost quadrupled to meet minimal standards. Such manpower shortages clearly have negative effects on equitable distribution of care; provision of comprehensive,

9. David Witt, MD, presented at The Third National Conference on Genetics and Public Health, September 20, 2000.



ongoing care management; and effective teaching and education.

**Issue 4: Lack of Adequate Funding.** As noted, inadequate reimbursement for certain genetic services has been well documented. This has created a continued dependence on public funding. In addition, State funding for genetic services has, allowing for inflation, decreased each year since 1989.<sup>10</sup> Non-self supporting services will be unable to continue if additional funding sources are not identified. Furthermore, there is no current mechanism to secure reimbursement for many of the services provided to patients by genetic counselors. Consequently there is limited public and private support for such counselor-based activities.

**Issue 5: Challenges of Providing Equitable Care and Access to Care.**

The Partnership for Genetic Services Pilot Program, sponsored by the Alliance of Genetic Support Groups (now the Genetic Alliance) identified the consumer indicators of quality genetic services (see Appendix V). The first identified priority was assurance that consumers will be referred to condition-specific specialists. However, inadequacies in the genetic workforce and the uneven distribution of manpower that does exist create a considerable challenge to assuring that all families in need will be appropriately referred. Fundamental barriers include those of availability and of access. In addition, a series of further obstacles may conspire to limit access to genetic services. These obstacles include:

- Physicians and other primary care providers who are unaware of the need and appropriateness of genetic referrals;
- Lack of awareness by other health care providers (such as public health nurses, social workers etc.) of the need for genetic services;
- Lack of consumer awareness and understanding regarding genetic services;
- Barriers imposed by third-party payers, special barriers for those who are uninsured or underinsured, and other structural barriers to access—
  - In 1998, 4% of Wisconsin residents had no health insurance of any kind in the previous 12 months and an additional 6% had health insurance for only part of the year;

- Individuals who are members of minority groups, had less than a high school education and/or are in poverty are most likely to be uninsured;<sup>11</sup>
- Geography, since services that might be readily available in Milwaukee or Madison, for example, may be far more difficult to access in outlying regions;
- Sociocultural barriers, including economic and language barriers;
- Attitudes and beliefs about genetic services, such as:
  - Lack of appreciation of the value of anticipatory medical care;
  - Issues of guilt, fear and other emotional barriers to seeking genetic services;
  - Inaccurate perceptions of the purposes of genetic services (e.g. that it is directive, that it is about abortion, that it is solely risk counseling, that it will inevitably lead to discrimination);
  - Myths about causes of birth defects and genetic testing.

**Issue 6: Need for Stronger Collaborations among Geneticists and with Other Health Care Providers.**

Currently, there is limited interaction between public and private genetic care providers. Furthermore, there is an overall lack of awareness of the benefits and availability of genetic services among other health care providers. Finally, with the development of new genetic knowledge and its clinical application, a greater need for increasing integration of genetics into all aspects of medical care will arise.

**Issue 7: Need for Additional Educational Activities,**

- including for consumers, for health and social service providers, for public policy makers and for payers. In order to maximize the benefits of genetic advances, educational activities need to continue to be offered at all levels—
- Continuing medical education of physicians
  - Postdoctoral training of physicians
  - Graduate training of genetic counselors
  - Teaching of medical students
  - Teaching of other health care providers
  - Continuing education of other health care providers
  - Teaching of educators at all levels
  - Informing the public
  - Informing families, support groups and other family-centered programs.

10. Current sources of funding for genetic services in the public sector are summarized in Appendix IV.

11. 1998 Family Health Survey, Wisconsin Bureau of Health Information.



## PERCEIVED WEAKNESSES AND CHALLENGES II: CHALLENGES FOR THE FUTURE

Two issues of singular importance will have extraordinary effects on medicine in the new millennium: the explosion of new genetic knowledge and changes in health care delivery. These issues and several others will have

marked consequences for clinical genetic services. From each issue, certain barriers to effective genetic service provision may arise. Because this document is intended to act as a guide to future actions, challenges are emphasized. However, each potential barrier can be viewed as providing **opportunities** for recruiting new advocates, welding new partnerships, expanding the scope of services and, ultimately, improving genetic care.

### “Epochs in the Genetic Understanding of Medical Disorders”

Clinical genetic services received scant attention in the first half of the past century. Beginning in the 1950s and 1960s substantial progress was made in the description and delineation of different causes of birth defects. This descriptive phase was critical not only from the perspective of providing care and counsel, but also as a prelude to the effective use of molecular technologies.

The 1960s and 1970s was the Epoch of Formal Mendelism as well as the time of the initiation of clinical care programs throughout the country.

The 1980s and 1990s ushered in what might be termed the Epoch of Molecular Mendelism. Using newly discovered techniques, researchers avidly hunted down genes that cause genetically straightforward processes. So, for example, we now know the genetic causes underlying the majority of instances of prelingual hearing loss, and know that one gene (GJB2 that codes for a product called connexin 26) is responsible for a major portion of genetic forms of deafness. Although incredibly important, the discoveries of the genes giving rise to such single gene processes are finite. In the very near future, all such loci will have been described.

We are currently experiencing the Epoch of the Molecular Delineation of Complex Disorders. That is, those same molecular techniques and the same knowledge gained from the Human Genome Projects are being applied to knottier questions regarding disorders that are sometimes clearly genetic and sometimes not, such as breast cancer and the discoveries related to the breast cancer predisposing genes, BRCA1 and BRCA2.

We are, it seems, early into another such epoch, one that might be called the Epoch of Molecular Understanding of Predispositions. That is, researchers are now searching for the complex patterns of normal, inherited variations to identify which of these are important in each of our unique predispositions to what are usually thought of as environmentally caused diseases. That is, in time (and probably a rather short time) it may be possible to identify the genomic “profile” in each of us – what we should

do to avoid disorders for which we have special risk, which lifestyle risk factors we can safely ignore, etc. If successful, this will lead to an explosion of diagnostic tests, screening packages and similar products. Not only will our knowledge about our own health risks explode, but therapeutics will likely be irrevocably altered as well. Pharmacogenomics (tailoring drugs to the particular genetic characteristics of the patient) holds considerable promise, but like all endeavors that are based on molecular genetics, also means that physicians, other health care providers, and all of us will need to become considerably more knowledgeable in order to use such information wisely. Gene therapy may also offer new promise of the treatment of disease in ways not previously possible.

Substantial unanswered questions accompany the molecular revolution that has overtaken genetics and the genetics revolution that is changing the face of medical care:

- Where do the resources for all of this testing come from?
- Who pays for such testing?
- Who provides the needed education of health care providers and how?
- How will access to counseling services be assured when demand may increase exponentially?
- When will treatment options become comparable with our diagnostic capabilities?
- How will we address the possible psychological consequences of testing – the effects of knowing “too much”, of having a “positive” test even at time of normal health, of having a “negative” test and experiencing so-called survivor guilt?
- How will society control the possible practical consequences of testing – in the workplace, on insurers?
- Will coercion (either tacit or explicit) to utilize available tests arise? Will coercion to act on abnormal test results occur?
- Will our society’s attitudes toward those with differences change?

# LIFE l i n e

## The New Genetics

"Mom, what type of breast cancer did Aunt Barb die from?" It was my daughter Sue on the phone. I could tell by the tone of her voice that it was no casual question. When I asked her why she was asking, her voice trembled, "I found a lump in my breast."

That was eight years ago and, at the time, my family knew little about the genetics of breast cancer. We just knew there seemed to be a "family link." It wasn't ever talked about much, but several of my aunts died from breast cancer. Barb, my sister, was diagnosed with the disease at age 39 and died at age 44. Now, my daughter too? Despite our family history, Sue's doctors told her that she was too young at the age of 33 for mammography. Eventually we convinced the doctors that mammography and a biopsy were needed. Sue had cancer.

My involvement with genetics started several years later when my primary physician cut my dose of Premarin, an estrogen replacement therapy drug. When I started having more and more severe hot flashes with the decreased dose, I went back to my doctor. She told me estrogen replacement therapy may be associated with an increased risk for breast cancer and because my daughter had breast cancer, she could not in good faith increase my dose without my seeing a genetic counselor. Soon after, I met Peter.

My family and I are lucky to have a good genetic counselor like Peter available. He has spent so much time with us, helping us to learn more about breast cancer and genetics, to understand our family history and our individual risks, and to think about the implications of genetic testing. After meeting with Peter, I decided to go through gene testing for breast cancer and yes, I was found to have changes in a breast cancer gene. I was prepared for this, not only because Peter had talked with me about the possibility, but also because my sister and daughter both had breast cancer. I guess I already assumed that I had to be "the link". Peter helped me to understand that having changes in this gene means that I am at increased risk for getting breast cancer (not that I will definitely get breast cancer). What next? Peter talked at length with me about options. I decided to have sur-

gery—a double mastectomy and oophorectomy.

As with any major surgery, I knew there would be risks. But at the time, I was healthy. I didn't have cancer. If I waited until I was sick with cancer, then I also might have to face chemotherapy and/or radiation. To me, that was scarier than having the surgery.

Because of the "genetic" nature of this information, I felt I needed to contact other family members. With input from Peter, I sent letters to immediate family members and first cousins on my father's side of the family (the side where all the breast cancer was found). The person most impacted at the time I sent the letter was my sister Barb's daughter. My niece told me, "All of my adult life, I have been unable to impress on anyone the concerns I have about getting breast cancer and testing for it. No one wanted to do mammography because I was 'too young'." Finally someone would be willing to listen to her concerns and help her find out whether she had the same gene change that led to her mother's breast cancer. My niece was tested. She wasn't surprised when the results came back indicating that she too carried the gene change; she had, in her own mind, always assumed that she would. Surgery (a double mastectomy) followed. Since all of this, my niece has had more peace of mind than she has had ever since her mother's diagnosis. She knows she is not "home free," but she feels this information has helped her to do something to cut a major risk factor by a huge percentage.

Deciding to go through genetic testing is not easy. While test results may provide helpful information, there are other concerns—cost, insurance, discrimination. Different members of my family have made different decisions about whether to test or not to test. We know this is the way it should be . . . it's a very personal decision. It has been reassuring for all of us to have genetic counselors to call upon to discuss these issues.



**Issue 1: The New Genetics.** While extraordinarily exciting, new genetic technologies are of concern to genetic health care providers. Through the Human Genome Project, an explosion of information will become available over the next decade that will revolutionize not just clinical genetics but all of medicine. A draft map of the human genetic makeup is complete. Current breakthroughs are mostly related to single gene disorders traditionally thought of as being genetic in origin. However, the genetic factors that play a role in common disorders such as various forms of cancer, heart disease, and asthma are being identified with increased frequency.

In the not so distant future, the potential for identification of genetic predisposition in a

much more global sense will be feasible. This will lead to the potential for widespread presymptomatic testing, far more extensive population screening, gene-specific strategies for therapy, and medical treatments tailor-made for the genetic makeup of the individual (i.e. pharmacogenomics). Why might these be the ingredients for a crisis? On the one hand there are far too few physician geneticists and genetic counselors to handle the needs that will arise. On the other, most physicians feel that they currently lack an adequate background in genetics to offer these services themselves.<sup>12</sup> Yet the impact that such possibilities may have on preventive health care is enormous.

The promise of the new genetics is manifold. There will be a vast increase in the ability

to make accurate diagnoses. Increased understanding of the causes of genetic disorders will lead to better treatment options. An explosion of beneficial population-based screening tools will be developed. Medication based on genetic information (pharmacogenomics) and gene therapy will likely become routine.

Challenges of this new paradigm of genetics in medical care may likewise prove to be massive. “*Genetics is everywhere*” will be truer than ever. Issues of an insufficient genetics workforce may prove overwhelming. Ongoing genetic education and training may require so much time from geneticists and genetic counselors that provision of direct services may suffer. In addition, a series of risks will need to be addressed. For example, how will the individual be protected given the potentials for discrimination (e.g. in insurance coverage) and loss of privacy (e.g. in the workplace)? Will third parties coerce individuals into genetic testing (either overtly or implicitly) and then discriminate based on the results? Will individuals opt for unneeded testing or rush to be tested without adequate consideration of the consequences?

## Issue 2: Changes of the Healthcare

**Marketplace.** The second major issue facing genetic service provision concerns health care financing and models of health care provision. The U.S. health care industry has become a far more competitive marketplace with increasing influence of various models of managed care. As a new specialty, Medical Genetics is problematic for many managed care organizations—how necessary are the services; are services cost-effective; what are the costs of “genetic benefits”? While it is easy for any medical specialty to claim that the need for its services is self-evident, it is likewise easy for managed care organizations to dismiss this assertion as self-serving. Furthermore, many medical directors have little experience with the role of geneticists in the health care delivery system and have very little formal training in genetics. Such factors may contribute to a series of issues that make provision of comprehensive genetic services within managed care organizations a greater challenge:

- In a cost-conscious setting, genetics may be seen by some to be an expendable luxury. The care of traditional genetic disorders is costly, resulting in more than a third of pediatric hospital admissions<sup>13</sup> and about

10% of adult admissions. In addition, individuals with genetic disorders often require outpatient care by multiple subspecialists.

- The first priority of consumers of genetic care is the ability to be referred to condition-specific specialists, even if out-of-plan (see Appendix V). However, few, if any, managed care organizations are large enough to support “supersubspecialists” (i.e. geneticists with special expertise in a particular group of genetic disorders).
- Many of the benefits of genetic care are not easily evaluated by evidence-based criteria. Subtle yet important benefits such as change in attitude, development of autonomous decision-making, alteration of reproductive choices, and impact on the medical care of extended family members do not conform to typical cost-effectiveness assessments.<sup>14</sup> Not all benefits of genetic care result in measurable changes in outcome, but absence of any change of action should not be viewed as a failed interaction.
- Since traditional genetic disorders are individually rare, it is exceedingly challenging to develop evidence-based care guidelines for each of them. This may become a greater issue in the future if supersubspecialists, who see and care for relatively large num-

## A Vision for the Future

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**Implementation of the principles identified in this plan will result in the seamless integration of genetic knowledge and care across the continuum of medical care delivery in Wisconsin. This will be accomplished through establishing new collaborations and alliances, providing ongoing education of all care providers, and crafting new approaches to the organization and funding of genetic services.**

...

13. Hall JG, Powers EK, McIvaine RT, Ean VH: The frequency and financial burden of genetic disease in a pediatric hospital. *Am J Med Genet* 1:17–36, 1978; Cassidy SB, Brunger JW, Moussavand S, McCandless SE: Now more than ever: The burden of genetic disease in over 4,000 consecutive pediatric hospital admissions.

American Society of Human Genetics National Convention, Philadelphia, 2000.



- bers of individuals with a particular rare disorder, are no longer available.
- Some models of managed care do not focus adequate resources on education, training and a public health orientation, all of which are, and should be, central to genetic care in its broader sense.

Despite these problems, many partnerships exist between geneticists and managed care organizations which are mutually beneficial and should be encouraged. Areas of potential cooperation and collaboration include issues of quality of care, access to appropriate health care providers, technology assessment, patient satisfaction and patient and provider education.

**Issue 3: Transitions to adulthood.** A third challenge of the future relates to a change in the focus of genetic care provision.

Historically, physicians trained in pediatrics and supported by pediatric focused funds have provided most clinical genetic care. Such an emphasis was appropriate since the greatest needs were present in children. However, many children with severe birth defects who in the past would have succumbed to their disorder, are now surviving into adulthood. Who will continue to provide comprehensive care to these individuals as they more frequently reach adulthood? Affected individuals

and their families will need to establish collaborations with both public and private service planners in order to assure continuing comprehensive services during this transition.

**Issue 4: Adult-onset disorders.** The new genetics that promises to provide information about predispositions to adult-onset diseases also implies a far greater emphasis on adult genetics than is currently the case. As this occurs, modifications of education, training and funding will need to more adequately be addressed. Some experience has accumulated regarding provision of genetic services to individuals at risk for certain adult-onset diseases. For example, providers and consumers in cancer genetics programs have experience that may assist in planning models of care for other adult-onset disorders.

GREEN BAY PRESS/SCIENCE



Ms. Ghate meets with a patient who has a family history of cancer.

# RECOMMENDATIONS OF THE WORKGROUP<sup>15</sup>

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The workgroup quickly recognized that a large number of issues face genetic services in the future. It had neither the resources nor time to find solutions to all of the problems it identified. Instead, it elected to make this summary one of needs identification rather than solutions. The workgroup identified a number of issues that need to be addressed and, in this section, suggests directions, approaches, and possible key players to finding solutions.

## CLUSTER 1: ADVISORY COUNCIL

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In order to continue its work the workgroup recommends that **a standing Advisory Council for Genetic Services be established through legislative mandate and modeled after the Newborn Screening Advisory Group and Council on Birth Defects Prevention and Surveillance.** It should have broad representation, including various care providers, State representatives, consumers, support organizations and other interested parties. **This Advisory Council for Genetic Services should be charged with continuing the assessment of the status of genetics and providing guidance regarding all of the following recommendations.**

**KEY PLAYERS:** State Plan Workgroup, Newborn Screening Advisory Group, Council on Birth Defects Prevention and Surveillance, Genetic Care Providers, Non-genetic Care Providers, Consumers, Consumer Advocacy Groups, Healthcare Payers, Department of Health and Family Services, Legislators, Local Public Health Departments

## CLUSTER 2: STATE PRESENCE

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Currently the only position in the Wisconsin Division of Public Health devoted to genetics is the State Genetic Coordinator. This position is located in the Bureau of Family and Community Health and is funded with monies from the Newborn Screening Program. While of great value, a single position, lacking in visibility and influence, is insufficient. Positions that are devoted to genetics are needed to advocate for genetics at the State level, especially related to funding, but also with respect to:

- education;
- grant administration;
- screening programs;
- prevention;
- serving as a resource regarding genetic issues in public health;
- conducting needs assessments;
- program monitoring;
- evaluation;
- overseeing genetic competencies within public health initiatives (comparable to Turning Point—the State Health Plan).

Although in the short term it is not realistic to change the placement of genetics in the state public health organization, ultimately **genetics in public health needs to be a program with more than one position at the State level and with greater autonomy and influence than exists currently.**

This program would be charged with coordinating and monitoring all genetic services in the State, overseeing the transition of emphasis to include adult disorders and disease predisposition, and providing liaison to existing public health groups. Its placement within the Division of Public Health should be the one to enhance connections with other relevant areas of public health. Within the newly established program, and in addition to genetic professionals, serious consideration should be given to the **creation of a position for consumer/patient liaison.** Strong consideration should also be given to establishing a State genetic epidemiologist position.

**KEY PLAYERS:** Department of Health and Family Services, Genetic Care Providers, Consumers, Consumer Advocacy Groups

## CLUSTER 3: SERVICE ISSUES—ASSESSMENT AND QUALITY ASSURANCE

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The Advisory Council should devise means of assessing current genetic services including:

- **Developing a State definition of minimum standards for clinics providing genetic services;**
- **Establishing means of licensing genetic counselors and certifying centers that provide genetic care;**
- **Considering the need and subsequently what mechanisms might be used to sim-**

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15. A summary of proposed activities for each key player is presented in Appendix VI.



- **ilarily identify, assess and certify comprehensive prenatal diagnostic centers;**
- **Reviewing published cost-effectiveness data regarding genetic care and devising ways of disseminating this information to the broader community;**
- **Generating quality assessment and care guidelines for such selected disorders;**
- **Reviewing published guidelines for care and reimbursement in the context of genetic services in Wisconsin.** These include, for example, information from official guidelines of the American College of Medical Genetics, the American Society of Human Genetics and the National Society of Genetic Counselors; Secretary's Advisory Committee on Genetic Testing guidelines; Hayes Technical Assessments; Aetna Coverage Policy Bulletins;
- **Developing quality assessment models for selected disorders and devising ways in which quality assessment information can be generated.** Examples of disorders that may be initially amenable to such an assessment include sickle cell disease, genetic testing for breast and ovarian cancer, stillbirth assessment, and teratogen information services;
- **Exploring alternative means of assessing quality of services.** These could include structured assessment of satisfaction with care, measures of changes in knowledge, measures of changes in perceptions related to genetic counseling and others;
- **Developing a position statement to address the difficulties of establishing standards of care for rare disorders;**
- **Assuring that consumers and their families are involved in all levels of assessing services and establishing guidelines for care.**

**KEY PLAYERS:** Department of Health and Family Services, Genetic Care Providers, Consumers, Consumer Advocacy Groups, National Organizations (e.g. American College of Medical Genetics, American Society of Human Genetics, National Society of Genetic Counselors, etc.), Department of Regulation and Licensing, Healthcare Payers

**CLUSTER 4: SERVICE ISSUES—INSUFFICIENT GENETICS WORKFORCE**

In collaboration with the Wisconsin Birth Defects Prevention and Surveillance Program and others involved in collecting epidemio-

logic data that concern genetic processes, **the Advisory Council should generate a needs assessment methodology and provide recommendations regarding manpower needs in genetics within the State.** Essential activities within this cluster include:

- **Taking an accurate census of genetic care providers and a measure of their activities;**
- **Providing an estimate of current and future needs based on available data from sources such as the Kaiser Permanente assessment;**
- **Identifying means of encouraging genetic training programs within the State in order to meet the anticipated needs in the future.**

**KEY PLAYERS:** Department of Health and Family Services, Genetic Care Providers, University-Based Training Programs, Healthcare Payers

**CLUSTER 5: SERVICE ISSUES—FINANCING GENETIC SERVICES**

Without additional public or private funding, access to genetic services will be limited and the ability of genetic providers to respond to the explosion of new information and new options will be severely hampered. What should be the State's financial commitment? What options for additional service funding should be pursued? **The Advisory Council should explore options for funding additional services and should review methods used in other States. In addition, it should:**

- **Seek a legislative mandate assuring adequate reimbursement of all types of genetic services;**
- **Identify and seek implementation of methods to allow billing for genetic counselor services;**
- **Identify possible additional and alternative public or private sources of funding for genetic services;**
- **Assess financial obstacles to accessing newly available genetic services, including issues related to reimbursement for molecular testing, coverage of testing of family members, etc.;**
- **In collaboration with third party pay-**

ers, establish criteria and priorities for services to be provided and identify priority target groups for such service provision.

**KEY PLAYERS:** Department of Health and Family Services, Genetic Care Providers, Consumers, Healthcare Payers, Department of Regulation and Licensing, Division of Health Care Financing, Medicare/Medicaid, Legislators

## CLUSTER 6: SERVICE ISSUES—ACCESS

It is imperative that provision of genetic care in Wisconsin is appropriate, comprehensive, and longitudinal regardless of health insurance status. Primary health care providers need to appropriately make available referrals for consultation with genetics professionals. In order to facilitate this, **the Advisory Council should develop a position statement regarding the special features of genetic services and the need to assure access to diagnosis-specific genetics experts. In addition, this strategic plan and subsequent materials generated by the Advisory Council should be widely distributed to both health care providers and payers.**

**A handbook of available genetic services in Wisconsin based on recognized quality standards (see Cluster 3) should be developed, published and distributed. Consideration should be given to sponsoring of a “genetic health summit” to discuss:**

- **access to genetic care;**
- **perceived barriers to access;**
- **insurance and managed care issues;**
- **issues of confidentiality and discrimination.**

One option that may improve access regardless of geography is telemedicine. However, providing long distance care in this manner presents considerable obstacles. **The Advisory Council should explore telemedicine options, particularly with regard to issues of legality, practicality, effectiveness and reimbursement.**

**KEY PLAYERS:** Department of Health and Family Services, Children with Special Health Care Needs (CSHCN) Regional Centers, Genetic Care Providers, Non-genetic Care Providers, Consumers, Consumer Advocacy Groups, Healthcare Payers, Local Hospitals and Clinics, Wisconsin State Medical Society, Wisconsin Hospital Association

## CLUSTER 7—COLLABORATIONS

Although collegial interactions prevail among most clinical geneticists in Wisconsin, it is the sense of this workgroup that improved collaborations are both desirable and feasible. Mechanisms that may be explored include

- **Formalizing relationships among geneticists through existing organizations such as the Greater Wisconsin Genetics Exchange and the Wisconsin Genetic Counselors Committee;**
- **Exploring means of forging closer collaborative ties among clinical geneticists, laboratory services and prenatal diagnosticians;**
- **Exploring means of establishing collaborative activities between managed care organizations and genetic care providers, particularly regarding access, cost and consequence assessments, technology assessment and educational activities;**
- **Exploring methods of establishing closer ties between providers of care and families, support groups and advocacy organizations.** Families, support groups, and advocacy organizations may prove pivotal in establishing the impetus for collaboration between geneticists and other specialties mentioned above.

**KEY PLAYERS:** Genetic Care Providers, Non-genetic Care Providers, Healthcare Payers, Consumers, Consumer Advocacy Groups

## CLUSTER 8—ISSUES RELATED TO THE NEW GENETICS

As already emphasized, new genetic technology and new diagnostic methods will not only change the face of genetic services, but also fundamentally change the practice of medicine in general. For genetics, the most pressing issues will be related to adequate education of primary care practitioners and to genetics workforce limitations. Recommendations to be addressed by the Advisory Council include:

- **Generating recommendations for primary care education related to molecular diagnostics;**
- **Exploring alternative counseling models and generation of recommendations regarding their use;**
- **Developing and monitoring programs**

**that ensure that genetic tests and services are integrated into population-based interventions that promote health and prevent disease and disability.**

**KEY PLAYERS:** Department of Health and Family Services, Genetic Care Providers, Non-genetic Care Providers, Consumers, Consumer Advocacy Groups, Educators, Healthcare Payers

### CLUSTER 9—EDUCATION RELATED ISSUES

The workgroup is convinced that educational initiatives are fundamentally important to the future viability of genetic services. Educational efforts should be directed towards physicians, other health professionals, administrators, State personnel, legislators, the public and those in need of direct genetic services. Initiatives should include:

- **Creating a Statewide Genetics Website including subsections for professionals and for consumers/the public.** It should include linkages to sites of relevance to genetic care;



Theresa Shuck, a genetic counselor at St. Vincent's Hospital in Green Bay, meets with a patient.

- **Developing a plan to expand educational programs, including recommendations for funding such activities.**

Specific activities should include:

- Exploring possible partnerships with the March of Dimes, Wisconsin Association for Perinatal Care, Wisconsin Public Health Association, consumer and support groups, and others;
- Developing and maintaining a speakers bureau;

- Developing an 'experts list' of both geneticists and consumers for the media;
- **Identifying alternative educational strategies and of methods for funding of unique or experimental educational approaches;**
- **Exploring creation of a Wisconsin Organization for Rare Disorders;**
- **Exploring methods of improving educational opportunities for**
  - Medical students and residents in all specialties;
  - Practicing physicians (particularly related to new genetic technologies);
  - Nurses, nurse practitioners, physician's assistants and therapists;
  - The public, including publicizing the existence of:
    - > Available resources, speakers, etc.
    - > Genetic Alliance, Ambassadors for Awareness etc.
    - > Legislators and other public officials;
- **Surveying current requirements for genetic education of each healthcare professional group trained in the state and generating a position statement on the need for such training.**<sup>16</sup>

**KEY PLAYERS:** Department of Health and Family Services, CSHCN Regional Centers, Genetic Care Providers, Non-genetic Care Providers, Consumers, Consumer Advocacy Groups, Educators, Universities, Medical Schools, Schools of Allied Health, Healthcare Payers, March of Dimes, Wisconsin Association for Perinatal Care, Wisconsin Public Health Association, Genetic Alliance, National Organizations (e.g. ACMG, ASHG, NSGC, etc.), National Coalition for Health Professional Education in Genetics, Local Hospitals and Clinics, Wisconsin State Medical Society, Media

Finally, among its other activities, the Advisory Council may function as a primary source of information dissemination about new tests, new methods of assessment and other issues of relevance to the larger community.

We fully recognize that these recommendations are too numerous and too challenging to be addressed simultaneously. **The Advisory Council will have, as its first task, further prioritizing these needs.**

16. Core Competency Working Group of the National Coalition for Health Professional Education in Genetics: Recommendation of core competencies in genetics essential for all health professionals. *Genetics in Medicine* 3:155-159, 2001.



# APPENDIX I

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## WORKGROUP ON DEVELOPMENT OF A GENETIC SERVICES PLAN FOR WISCONSIN

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## APPENDIX II

### GENETIC SERVICES AND THE COUNCIL OF REGIONAL NETWORKS (CORN) GUIDELINES

Recently CORN developed a set of comprehensive Guidelines for Clinical Genetic Services for the Public's Health<sup>1</sup>. Given that the former director of this State's largest clinical genetic center, Dr. Renata Laxova, had primary responsibility for drafting these guidelines, it is not surprising that genetic services in Wisconsin closely reflect the recommendations made there. Nevertheless, while services as a whole conform to the recommendations of the CORN guidelines, clearly substantive deficiencies exist. Likewise, certain of those deficiencies will reach critical proportion because of the issues discussed above.

The following highlights areas of strength and weakness in genetic services in Wisconsin with respect to the CORN guidelines.

#### Strengths of Clinical Genetics in Wisconsin

##### I. Organization and Administration

###### I.A State/Territorial Genetic Coordinator/Educator

A genetic counselor serves within the State system as genetic coordinator. Most (but not all) of the recommended activities within this guideline are currently being carried out.

###### I.D.1 Structure of the State/Territorial Genetic Services Network

Through the efforts of many providers, various models of care are extant within the network of genetic services. Indeed, as the University-based clinical genetic program developed, it was predicated on the assumption that outreach activities, in particular, would evolve through greater and greater levels of sophistication and autonomy. Just such an evolution has occurred. Currently in this State there are two, large comprehensive University-based genetic centers, two genetics units within managed health care facilities, seven outreach genetics sites, as well as approximately five private, for-profit independent provider groups.

###### I.D.2 Types of Services

Within those that are University-based or University-affiliated, considerable emphasis is placed on providing family centered care. In addition, active screening programs are extant including: prenatal screening (available through the State Laboratory of Hygiene); newborn screening (through the State Laboratory of Hygiene) and follow-up (coordinated by the State and provided in the Biochemical Genetics Clinic and through other providers); birth defects monitoring, through the newly established Program and Council on Birth Defects Prevention and Surveillance; teratogen information services, with a large centralized service in Madison, and smaller pro-

grams in Milwaukee, La Crosse and Green Bay; a statewide community based stillbirth assessment program.

In addition, there is an extensive educational effort coordinated for the most part through the University Clinical Genetic Center providing outreach education to medical professionals, other professionals (e.g. teachers) and for the public.

###### I.E.1 Assurance

Formal policy of the Clinical Genetic Center of the University and of Children's Hospital of Wisconsin assures access regarding all listed attributes. The State has little control over access to other providers.

###### I.E.2 Education

As noted, the University-based programs have placed great emphasis on community and consumer education.

###### I.E.3 Referral

While referral to providers in this State is straightforward, managed care impediments are not infrequently extraordinarily difficult to overcome. Development of an "efficient referral system" will require more than the good will of clinical geneticists.

###### I.E.4 Quality of Service

All University-affiliated programs in the State are appropriately compliant. All University-affiliated providers are Board certified or Board eligible.

###### I.E.5 Privacy and Confidentiality. Compliant.

###### I.E.6 Ethical and Legal Standards. Compliant.

##### II. Prevention

###### A. Primary Prevention

###### II.A.1 Folic Acid Prevention of Neural Tube Defects

The State of Wisconsin, in collaboration with the March of Dimes and genetic providers undertook a folic acid education campaign and folic acid alert. There has been no direct measure of its impact, however.

###### II.A.2 Prenatal Exposures

As noted a central Teratogen Information Service is available at the University of Wisconsin-Madison, with additional informational sites in Milwaukee, La Crosse, and Green Bay.

###### II.A.3 Maternal Disease Management

Specific collaborations with high-risk obstetricians are in place but such collaborations are in no way codified or formalized.

###### II.A.4 Preconceptional Counseling

Preconceptional counseling is, in general, readily available (given the universal difficulty of successful referral related to third party payers).

###### II.A.5 Adult Onset Disorder Counseling

Cancer risk counseling, in particular, has expanded extraordinarily rapidly. In Madison a large effort, in collaboration with Comprehensive Cancer Center, is in place. Clinics devoted to such counseling also are now available in Milwaukee and La Crosse. Other adult-onset disorders have not yet had specific pro-

grams developed.

## B. Secondary Prevention

### II.B.1 Newborn Screening

The State's newborn screening program is well established with comprehensive follow-up and oversight by a Newborn Screening Advisory Group. This state has been a leader in assessing and incorporating additional disorders into the newborn screening program.

### II.B.2 Prenatal Screening

Prenatal screening is provided, for the most part, through private providers. Virtually by definition this means that coordination is generally lacking and access spotty.

### II.B.3 Other Screening

Selected screening programs are currently extant including ones for cancer and for Huntington disease, for example.

## C. Tertiary Prevention

In general, University-based clinics in this State have emphasized, more than most, ongoing management and care (as opposed to being primarily or solely diagnostic clinics) of individuals with very specialized health care needs.

## III. Services

### A. Types of Service

#### III.A.1 Family Focused Services

All sites throughout the State have the equivalent of general genetic clinics. The primary biochemical genetic clinics are located in Madison and Milwaukee. Single disease programs are found throughout the State, including, for example, sickle cell programs in Milwaukee, craniofacial disorders clinics in Madison, Milwaukee and La Crosse, the Comprehensive Cancer Center in Madison etc. Prenatal clinics are, for the most part, in the private sector. Close collaboration exists between the two prenatal diagnosis centers in Madison and the Clinical Genetic Center there.

#### III.A.2 Population Oriented Services

All screening programs listed are currently available.

#### III.A.3 Clinical Laboratory Services

In addition to private cytogenetics laboratories, two are affiliated with the University of Wisconsin. A central Biochemical Genetic Laboratory is also located there. Selected molecular diagnoses are carried in the Molecular Genetic Laboratory of the University of Wisconsin, and elsewhere, including by various private care providers.

### III.C Facility Requirements

The public providers are compliant for all guidelines provided including those related to physical facilities, access, accreditation, cultural appropriateness, quality assurance, consumer involvement, administrative organization, contractual arrangements, protocols, billing policies etc.

### III.D Staffing and Credentialing

There are currently approximately 17 Board certified M.D. geneticists and 29 genetic counselors in the State, as well as individuals within all of the other specialty categories listed. All of those affiliated with University-based programs are appropriately certified.

### III.E Components of a Genetic Evaluation

All of the activities of University-based programs are fully compliant with these recommendations.

### III.F Patient Records

All of the activities of University-based programs are fully compliant with these recommendations.

### III.G Human and Legal Rights

All of the activities of University-based programs are fully compliant with these recommendations.

### III.H Quality Assurance

All of the activities of University-based programs are fully compliant with these recommendations.

## IV. Research

Active collaborative clinical research is ongoing. All research activities are aware of issues of informed consent, confidentiality, risk etc. and all research activities are evaluated and approved by appropriate Protection of Human Subjects Committees (Institutional Review Boards).

## V. Education

Education and training have traditionally been strengths of Genetic programs in this State. They remain so. Such activities include hundreds of individual presentations to thousands of consumers, physicians, teachers and others each year. They include special ongoing programs such as Summer Teacher Enhancement Workshops, Primary Care Education in Genetics programs etc.

### Previous Efforts to Address Needs and Deficiencies

Using the CORN Guidelines, two groups were constituted with the leadership of the University of Wisconsin Clinical Genetic Center and the State Genetic Coordinator in 1997–1998. Termed "The Future of Genetics in Wisconsin: Vision for Care", and "The Future of Genetics in Wisconsin: Prospects for Funding". These groups grappled with issues already summarized. They identified a list of problems that genetic care provision needed to face. Most related to the issues of the impact of the molecular genetic revolution, manpower, funding, relationships with managed care providers and needs for further grass roots education about the importance of genetic services. These initial discussions form the basis for the far more extensive consideration developed here.

1 Guidelines for Clinical Genetic Services for the Public's Health, 1st edition. Council of Regional Networks for Genetic Services, 1997. Pp. 13–33.

## Areas of Apparent Deficiency

In addition to two major issues addressed in the main body of this document – challenges derived from changes in health care delivery and concerns regarding the explosion of genetic knowledge still to come – certain more specific deficiencies are apparent based on comparisons with the CORN guidelines. These include the following.

### I.B State/Territorial Advisory Council

Wisconsin has no Genetic Advisory Council. Obviously, it is hoped that this plan will be the initial step in creating such a council.

### I.C State/Territorial Plan for Genetics

While Wisconsin is rich in tradition, has a high level of clinical genetic services and extensive outreach and education programs in place, no formal plan for provision of services has heretofore existed. This document rectifies this deficiency.

### II.A.3 Management of Maternal Diseases

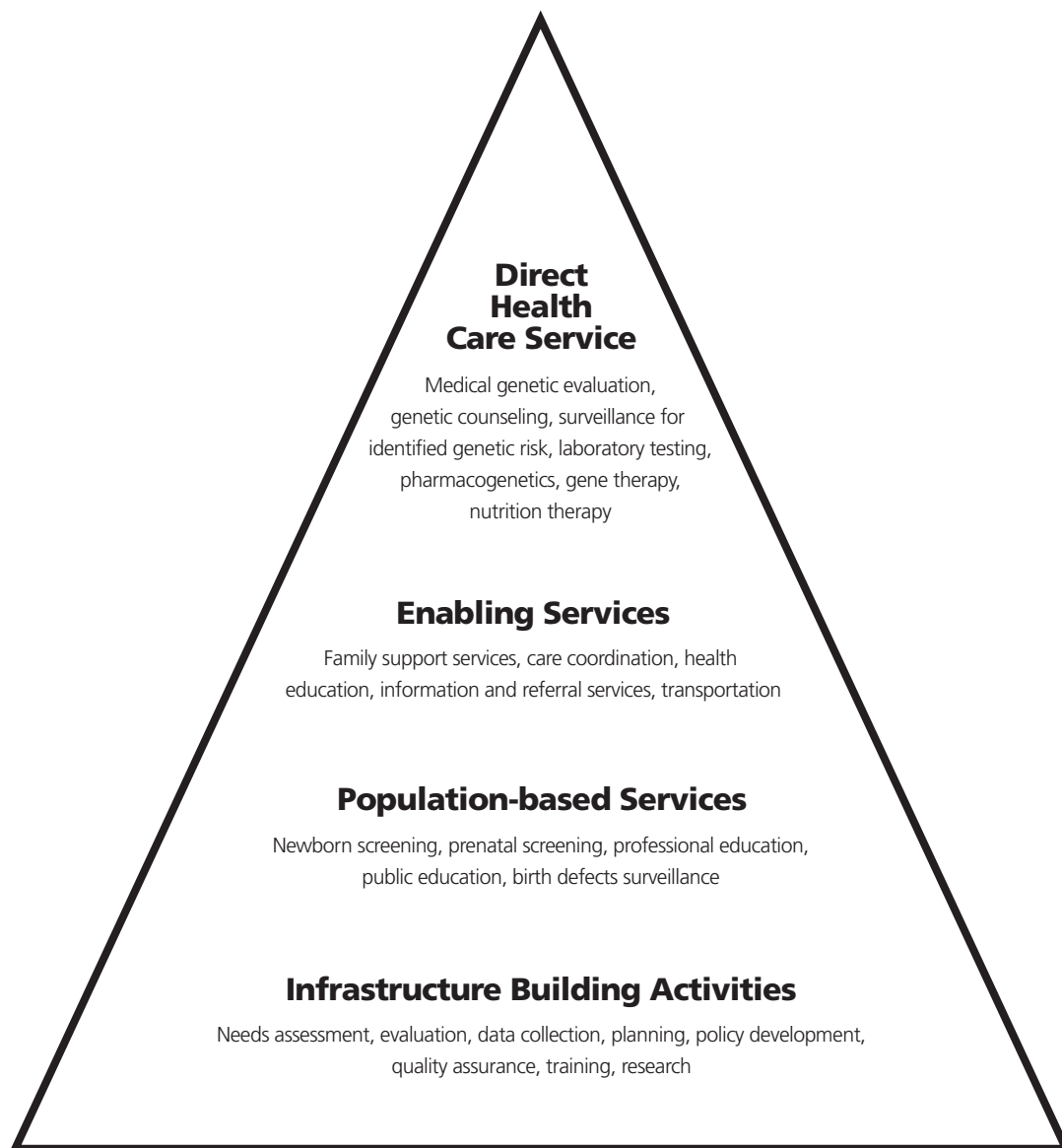
Collaborative relationships among geneticists and other providers are sometimes informal, tenuous and would be benefited from a more structured set of recommendations regarding how such collaborations should be maintained.



## APPENDIX III

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### GENETICS AND PUBLIC HEALTH PYRAMID



# APPENDIX IV

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## PUBLIC FUNDING FOR GENETIC SERVICES IN WISCONSIN

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### Overview

Funding for genetic services is complex, both here and elsewhere in the United States. Currently there is no national standard for funding and each state has its own mix of funding mechanisms. Furthermore, even within one state different genetics activities may be funded independently, and between states the same activity may be funded entirely differently. Funding sources include newborn screening fees, general-purpose revenue, pass through money from federal sources, University funds, fee for service collection, various private and public grants and contracts.

### Past Funding in Wisconsin

Initially clinical services were 'bootlegged' onto research funding. Monies specifically for the provision of genetics services were not received until 1976.

From 1976 to 1981 a statewide grant from the Developmental Disabilities Council of Wisconsin helped establish what was then termed the Genetic Contact Program. "Genetic Contacts" were individuals in each county of the state who were trained to recognize and refer individuals and families who were in need of clinical genetic services. Early prenatal diagnostic services also developed through money obtained in this grant.

The National Genetic Disease Act (Title XI of the Public Health Service Act) made federal funds available for genetic services for the first time. Grants were obtained for 1979 through 1982 for the development of a genetic services program in Wisconsin.

With the transformation of federal funding into a Block Grant program, Title V monies became competitively available. A portion of public sector genetics services has been funded with Title V monies for Statewide Genetic Services.

In addition, awards have previously been obtained from many private funding agencies for one aspect or another of genetics care. Medical schools in the State provide salary funding (including five physician geneticists).

### Current Funding in Wisconsin

Funding of public sector genetics currently includes a major grant for the Statewide Genetic Service Program (centered in Madison with subcontracts for LaCrosse, Eau Claire and Milwaukee) through the Department of Health and Family Services. In addition, certain genetic services (e.g. biochemical genetics) are funded through the Congenital Disorder Program that is supported by fees from newborn screening. Physician salaries are provided by the University of Wisconsin–Madison

(State 101 funded) and the Medical College of Wisconsin/Children's Hospital of Wisconsin.

In addition to these major sources of funding, maintaining a comprehensive program has required cobbling together a heterogeneous mix of funding. For example, over the past two years the Clinical Genetics Center at the University of Wisconsin has utilized 23 different funding sources including other units of the University, private foundations, contracts with private hospitals, biotechnology companies, various state agencies, federal agencies etc. Virtually all programs are dependent on 'soft' money for their continued existence.

### Funding Mechanisms in Neighboring States

Nationwide, state-specific information on funding for genetic services is not available. A state-by-state survey was recently completed under the direction of Hawaii's state genetic coordinator. However, state profiles are not yet available. Once completed, this survey may be of considerable benefit to the Advisory Council for Genetic Services.

Since state-specific information is not yet accessible, project staff carried out an informal survey to assess how genetic services are being funded in Wisconsin's neighboring states.

- **Iowa.** Iowa has general revenue funds dedicated to genetic services. These support the Regional Genetic Consultation Service (~\$567,000) and the Neuromuscular and Related Genetic Disorders Program (~\$115,000). In addition, the Neonatal Metabolic Screening Program and the Expanded MSAFP Screening Program generate revenues, some of which are used to support a State Genetic Coordinator position. Iowa is one of the few states in which general revenue funds support a majority of the genetic services available in the state.
- **Illinois.** Revenue generated from newborn screening currently funds all of the public sector genetic activities. These fees also support approximately \$1,000,000 in costs for costs of newborn screening follow-up (PKU formula, laboratory staff, clinic staff). Beginning in 2002 general revenue funds will, for the first time, become available for genetic services (\$275,000). It is anticipated that the level of General Purpose Revenue (GPR) will increase in subsequent years.
- **Michigan.** All public sector services are supported by funds generated from newborn screening fees.
- **Minnesota.** No dedicated public money is available for general genetics services. Services are funded through third party payers (both public and private) and some funds from the Minnesota Children with Special Health Care Needs program.



## APPENDIX V

### CONSUMER INDICATORS OF QUALITY GENETIC SERVICES

FROM THE GENETIC ALLIANCE WEBSITE  
[WWW.GENETICALLIANCE.ORG/ABOUTUS/PUBLICATIONS/CQIBROCH.HTML](http://WWW.GENETICALLIANCE.ORG/ABOUTUS/PUBLICATIONS/CQIBROCH.HTML)

**Order reflects consumer prioritization**

1. Consumers are referred to condition-specific specialists, as needed, including those out-of-plan.
2. Medications and supplements to treat genetic conditions are covered by the plan.
3. Consumers and providers collaborate on ways to identify service needs, develop and monitor treatment plans, and manage a genetic condition.
4. Resources are available to assist consumers and their families in understanding:
  - symptoms;
  - screening/testing options and their implications; and
  - a diagnosis.
5. Providers consider the medical and psychosocial impact of a genetic condition on both individuals and their families at each stage of life.
6. A practitioner, experienced in genetic services, is available to plan members.
7. Consumers may choose their providers.
8. Policies and procedures about confidentiality of genetic information are in place.
9. Information about genetic conditions is provided to each consumer in a manner best suited to their needs and culture, which may include:
  - their primary language;
  - an appropriate educational level; and
  - more than one medium.
10. Information about genetic research is available to the consumer and integrated in clinical practices.

11. Referrals to genetic peer support resources are offered at regular office visits.

12. Outpatient, home, and hospice care for individuals with genetic conditions is available.

The Genetic Alliance gives a voice to the common concerns of persons living with, or at risk for, genetic conditions and their families. An active coalition of consumers, professionals, genetic support groups and organizations, the Alliance builds partnerships to promote optimum healthcare and enhanced quality of life.

Representing over 300 support groups the Genetic Alliance links consumers, professionals and the public to genetic support groups and resource referrals through a toll-free helpline and website. The website offers a growing store of links to member webpages and their condition-specific resources—providing educational and support resources for both the professional and consumer communities.

# APPENDIX VI

## KEY PLAYERS AND RECOMMENDATIONS

| Key Player   | Cluster                                       | Activities   |   |
|--|---|--|---|
| Children with Special Health Care Needs Regional Centers | 6: Service—Access                             | <ul style="list-style-type: none"> <li>Disseminate Plan and subsequent materials</li> <li>Position paper on access</li> <li>Handbook of available genetic services in Wisconsin</li> <li>Sponsor ‘genetic health summit’</li> <li>Issues of telemedicine</li> </ul>  |   |
|  | 9: Education                                  | <ul style="list-style-type: none"> <li>Website creation</li> <li>Expansion of educational programs</li> <li>Identification of alternative strategies</li> <li>Consider W-ORD creation</li> <li>Identify methods of educational improvement</li> <li>Assess genetic education requirement</li> </ul>  |   |
| Consumer Advocacy Groups                                 | 1: Advisory Council Creation                  | <ul style="list-style-type: none"> <li>Creation of an advisory council for genetic services</li> </ul>   |   |
|  | 2: Increasing State Presence                  | <ul style="list-style-type: none"> <li>Development of a State <i>program</i> in genetics</li> <li>Create Consumer Liaison position</li> </ul>  |   |
|  | 3: Service – Assessment and Quality Assurance | <ul style="list-style-type: none"> <li>Define minimum standards for genetics centers</li> <li>Develop quality assessment models</li> <li>Generate quality assessment and care guidelines</li> <li>Review cost-consequence data</li> <li>Review published guidelines for care</li> <li>Review published guidelines for reimbursement</li> <li>Develop position statement re establishing standards of care</li> <li>Explore methods of assessing quality</li> <li>Insure licensing of genetic counselors</li> <li>Insure certifying care providing centers</li> </ul> |   |
|  | 6: Service—Access                             | <ul style="list-style-type: none"> <li>Disseminate Plan and subsequent materials</li> <li>Position paper on access</li> <li>Handbook of available genetic services in Wisconsin</li> <li>Sponsor ‘genetic health summit’</li> <li>Issues of telemedicine</li> </ul>  |   |
|  | 7: Service—Collaboration                      | <ul style="list-style-type: none"> <li>Formalize relationships</li> <li>Explore collaborations</li> </ul>  |   |
|  | 8: The New Genetics                           | <ul style="list-style-type: none"> <li>Recommendations for primary care education</li> <li>Explore alternative counseling models</li> <li>Develop assurance of integration of testing into health promotion</li> </ul>   |   |
|  | 9: Education                                  | <ul style="list-style-type: none"> <li>Website creation</li> <li>Expansion of educational programs</li> <li>Identification of alternative strategies</li> <li>Consider W-ORD creation</li> <li>Identify methods of educational improvement</li> <li>Assess genetic education requirement</li> </ul>  |   |
|  | Consumers                                     | 1: Advisory Council Creation   | <ul style="list-style-type: none"> <li>Creation of an advisory council for genetic services</li> </ul>  |
|  |   | 2: Increasing State Presence   | <ul style="list-style-type: none"> <li>Development of a State <i>program</i> in genetics</li> <li>Create Consumer Liaison position</li> </ul> |
| 3: Service – Assessment and Quality Assurance            |   | <ul style="list-style-type: none"> <li>Define minimum standards for genetics centers</li> <li>Develop quality assessment models</li> <li>Generate quality assessment and care guidelines</li> <li>Review cost-consequence data</li> <li>Review published guidelines for care</li> <li>Review published guidelines for reimbursement</li> <li>Develop position statement re establishing standards of care</li> <li>Explore methods of assessing quality</li> <li>Insure licensing of genetic counselors</li> <li>Insure certifying care providing centers</li> </ul> |   |
| 6: Service—Access  |   | <ul style="list-style-type: none"> <li>Disseminate Plan and subsequent materials</li> </ul>  |   |



## APPENDIX VI

| Key Player   | Cluster                                     | Activities   |
|--|---|--|
|  | 7: Service—Collaboration                    | <ul style="list-style-type: none"> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> </ul>   |
|  | 8: The New Genetics                         | <ul style="list-style-type: none"> <li>• Formalize relationships</li> <li>• Explore collaborations</li> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> </ul>  |
|  | 9: Education                                | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
| Council on Birth Defects Prevention and Surveillance | 1: Advisory Council Creation                | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> </ul>   |
| Department of Health and Family Services             | 1: Advisory Council Creation                | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> </ul>   |
|  | 2: Increasing State Presence                | <ul style="list-style-type: none"> <li>• Development of a State <i>program</i> in genetics</li> <li>• Create Consumer Liaison position</li> </ul>  |
|  | 3: Service—Assessment and Quality Assurance | <ul style="list-style-type: none"> <li>• Define minimum standards for genetics centers</li> <li>• Develop quality assessment models</li> <li>• Generate quality assessment and care guidelines</li> <li>• Review cost-consequence data</li> <li>• Review published guidelines for care</li> <li>• Review published guidelines for reimbursement</li> <li>• Develop position statement re establishing standards of care</li> <li>• Explore methods of assessing quality</li> <li>• Insure licensing of genetic counselors</li> <li>• Insure certifying care providing centers</li> </ul> |
|  | 4: Service—Insufficient Genetics Workforce  | <ul style="list-style-type: none"> <li>• Take census</li> <li>• Estimate current and future needs</li> <li>• Identify means to encourage training programs</li> </ul>  |
|  | 5: Service—Finance                          | <ul style="list-style-type: none"> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>   |
|  | 6: Service—Access                           | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> </ul>  |
|  | 8: The New Genetics                         | <ul style="list-style-type: none"> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> </ul>   |
|  | 9: Education                                | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
|  | Department of Regulation and Licensing      | 3: Service—Assessment and Quality Assurance  |



| Key Player  | Cluster  | Activities   |
|---|--|--|
|   | 5: Service—Finance   | <ul style="list-style-type: none"> <li>• Generate quality assessment and care guidelines</li> <li>• Review cost-consequence data</li> <li>• Review published guidelines for care</li> <li>• Review published guidelines for reimbursement</li> <li>• Develop position statement re establishing standards of care</li> <li>• Explore methods of assessing quality</li> <li>• Insure licensing of genetic counselors</li> <li>• Insure certifying care providing centers</li> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul> |
| Division of Healthcare Financing  | 5: Service—Finance   | <ul style="list-style-type: none"> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>   |
| Educational Institutions including: <ul style="list-style-type: none"> <li>• Medical Schools</li> <li>• Universities</li> <li>• Schools of Allied Health</li> </ul> | 9: Education   | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
| Educators   | 8: The New Genetics  | <ul style="list-style-type: none"> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> </ul>   |
|   | 9: Education   | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
| Genetic Alliance  | 9: Education   | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
| Genetics Care Providers   | 1: Advisory Council Creation<br>2: Increasing State Presence | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> <li>• Development of a State <i>program</i> in genetics</li> <li>• Create Consumer Liaison position</li> </ul>  |
|   | 3: Service – Assessment and Quality Assurance                | <ul style="list-style-type: none"> <li>• Define minimum standards for genetics centers</li> <li>• Develop quality assessment models</li> <li>• Generate quality assessment and care guidelines</li> <li>• Review cost-consequence data</li> <li>• Review published guidelines for care</li> <li>• Review published guidelines for reimbursement</li> <li>• Develop position statement re establishing standards of care</li> <li>• Explore methods of assessing quality</li> <li>• Insure licensing of genetic counselors</li> <li>• Insure certifying care providing centers</li> </ul>   |
|   | 4: Service—Insufficient Genetics Workforce                   | <ul style="list-style-type: none"> <li>• Take census</li> <li>• Estimate current and future needs</li> <li>• Identify means to encourage training programs</li> </ul>  |



## APPENDIX VI

| Key Player               | Cluster                  | Activities  |  |
|--------------------------|--------------------------|---|--|
|                          | 5: Service—Finance       | <ul style="list-style-type: none"> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>  |  |
|                          | 6: Service—Access        | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> </ul>                                   |  |
|                          | 7: Service—Collaboration | <ul style="list-style-type: none"> <li>• Formalize relationships</li> <li>• Explore collaborations</li> </ul>   |  |
|                          | 8: The New Genetics      | <ul style="list-style-type: none"> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> </ul>  |  |
|                          | 9: Education             | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul> |  |
|                          | Healthcare Payers        | 1: Advisory Council Creation  | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> </ul>   |
|                          |                          | 3: Service—Assessment and Quality Assurance   | <ul style="list-style-type: none"> <li>• Define minimum standards for genetics centers</li> <li>• Develop quality assessment models</li> <li>• Generate quality assessment and care guidelines</li> <li>• Review cost-consequence data</li> <li>• Review published guidelines for care</li> <li>• Review published guidelines for reimbursement</li> <li>• Develop position statement re establishing standards of care</li> <li>• Explore methods of assessing quality</li> <li>• Insure licensing of genetic counselors</li> <li>• Insure certifying care providing centers</li> </ul> |
|                          |                          | 4: Service—Insufficient Genetics Workforce  | <ul style="list-style-type: none"> <li>• Take census</li> <li>• Estimate current and future needs</li> <li>• Identify means to encourage training programs</li> </ul>  |
|                          |                          | 5: Service—Finance  | <ul style="list-style-type: none"> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>   |
| 6: Service—Access        |                          | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> </ul>                                   |  |
| 7: Service—Collaboration |                          | <ul style="list-style-type: none"> <li>• Formalize relationships</li> <li>• Explore collaborations</li> </ul>   |  |
| 8: The New Genetics      |                          | <ul style="list-style-type: none"> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> </ul>  |  |
| 9: Education             |                          | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul> |  |

| Key Player                                    | Cluster                      | Activities  |
|---|------------------------------|---|
| Healthcare Providers<br>- other               | 1: Advisory Council Creation | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> <li>• Formalize relationships</li> <li>• Explore collaborations</li> <li>• Recommendations for primary care education</li> <li>• Explore alternative counseling models</li> <li>• Develop assurance of integration of testing into health promotion</li> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul> |
|   | 6: Service—Access            |   |
|   | 7: Service – Collaboration   |   |
|   | 8: The New Genetics          |   |
|   | 9: Education                 |   |
| Legislators                                   | 1: Advisory Council Creation | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>  |
|   | 5: Service—Finance           |   |
| Local Hospitals<br>and Clinics                | 6: Service—Access            | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor 'genetic health summit'</li> <li>• Issues of telemedicine</li> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
|   | 9: Education                 |   |
| Local Public Health<br>Departments            | 1: Advisory Council Creation | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> </ul>  |
| March of Dimes                                | 9: Education                 | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>   |
| Media   | 9: Education                 | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>   |
| Medicare/Medicaid                             | 5: Service—Finance           | <ul style="list-style-type: none"> <li>• Assess options for funding</li> <li>• Seek legislative mandate re reimbursement</li> <li>• Assure genetic counselor billing</li> <li>• Identify alternative sources of funding</li> <li>• Assess financial obstacles</li> </ul>  |
| National Coalition<br>for Health Professional | 9: Education                 | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> </ul>   |



## APPENDIX VI

| Key Player  | Cluster   | Activities  |
|---|---|---|
| Education in Genetics   |   | <ul style="list-style-type: none"> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |
| National Organizations such as: <ul style="list-style-type: none"> <li>• American College of Medical Genetics (ACMG)</li> <li>• American Society of Human Genetics (ASHG)</li> <li>• National Society of Genetic Counselors (NSGC)</li> </ul> | 3: Service – Assessment and Quality Assurance<br><br>9: Education | <ul style="list-style-type: none"> <li>• Define minimum standards for genetics centers</li> <li>• Develop quality assessment models</li> <li>• Generate quality assessment and care guidelines</li> <li>• Review cost-consequence data</li> <li>• Review published guidelines for care</li> <li>• Review published guidelines for reimbursement</li> <li>• Develop position statement re establishing standards of care</li> <li>• Explore methods of assessing quality</li> <li>• Insure licensing of genetic counselors</li> <li>• Insure certifying care providing centers</li> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul> |
| Newborn Screening Advisory Group  | 1: Advisory Council Creation                                      | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> </ul>  |
| State Plan Workgroup  | 1: Advisory Council Creation<br>9: Education                      | <ul style="list-style-type: none"> <li>• Creation of an advisory council for genetic services</li> <li>• Wisconsin Association for Perinatal Care</li> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>   |
| Wisconsin Hospital Association  | 6: Service—Access   | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor ‘genetic health summit’</li> <li>• Issues of telemedicine</li> </ul>   |
| Wisconsin Public Health Association   | 9: Education  | <ul style="list-style-type: none"> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>   |
| Wisconsin State Medical Society   | 6: Service—Access<br>9: Education                                 | <ul style="list-style-type: none"> <li>• Disseminate Plan and subsequent materials</li> <li>• Position paper on access</li> <li>• Handbook of available genetic services in Wisconsin</li> <li>• Sponsor ‘genetic health summit’</li> <li>• Issues of telemedicine</li> <li>• Website creation</li> <li>• Expansion of educational programs</li> <li>• Identification of alternative strategies</li> <li>• Consider W-ORD creation</li> <li>• Identify methods of educational improvement</li> <li>• Assess genetic education requirement</li> </ul>  |