

# Have We Gotten Too Big For Our Genes?

And How Do We Get a Better Fit?



## Advocacy in Genetics

### A Teaching Guide and Workbook

(Special Edition with Supplemental Brochure on Genetics Services Delivery and Genetics Glossary)

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# About the Mountain States Genetics Network Consumer Issues Committee

The Mountain States Genetics Network (MSGN) was established in 1984 and is funded by a grant (#MCJ-081002-02) from the U.S. Department of Health and Human Services, Division of Maternal and Child Health, Genetic Diseases Services Branch.

The MSGN Consumer Issues Committee is one of several committees functioning under the MSGN umbrella. Its members bring consumer perspectives to the network's activities, facilitate consumer members' work within the network, and provide educational programs for consumers about genetics, new technologies, and legislative advocacy.

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The Advocacy Manual, the Guidelines for Genetics Services Delivery and the Glossary of Genetic Terms are available for viewing and printing on the Publications page of our website [www.mostgene.org](http://www.mostgene.org).

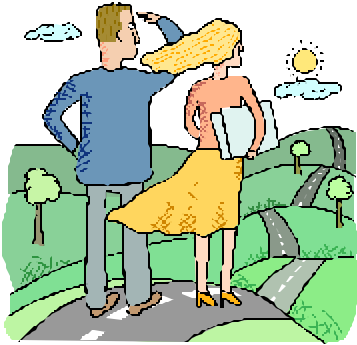
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## A Look into the Future through the Past



Human life and death used to be mysterious and uncontrollable events. Humans could not protect themselves from disease and dying. We believed that we were creatures with a destiny and that our destiny was inescapable.

Then scientists began to develop vaccines and antibiotics, and we could make ourselves better when we were sick. We could, with the help of science, avoid the plague of diseases that had once killed us. We could have an impact on our "destinies." And that was good.

Further technological developments, such as the x-ray and ultrasound, helped us to diagnose injuries and illnesses. Technology let us breathe when our respiratory systems failed. We had better tools to fight disease and death. And this was good.

Science and medicine continued to discover and refine our diagnostic methods and treatments. We were able to cure more and more once-devastating illnesses and injuries. And that was good.

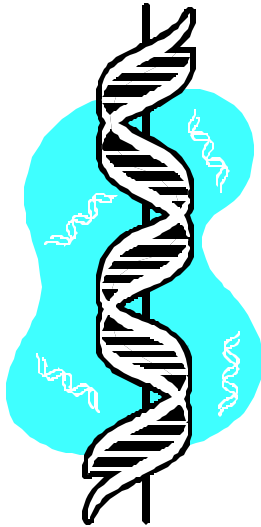
And then scientists set their sights on eliminating disease altogether. Why wait until a patient has leukemia to treat it; why not prevent it altogether? Why not use our medicine and technology before a disease started, instead of waiting until a person was already sick? And this was good.

The science of genetics now allows us to use carrier and prenatal testing to predetermine a child's predisposition for various diseases. And this is good.

Or is it? If we can control these elements in our lives, why not use this technology to control other things? And who would have access to use the technology? How do we, as a society, view the scope of disease and health; what would a world be like with less variability? Perhaps we could use this technology to ensure high IQs for all children, eliminating the need for special education services. The combination of genetic variables from which to pick and choose is infinite. Within the next few generations, the technology may exist to control some aspects of our own future – as well as that of our descendants. But what cost will we pay as a society and a species?

# The Human Genome Project

## The Human Genome Project And The Genetics Behind The Project: What Does It All Mean?



For all the diversity of the world's five and a half billion people, full of creativity and contradictions, the machinery of every human mind and body is built and run with fewer than 100,000 kinds of protein molecules. And for each of these proteins, we can imagine a single corresponding gene (though there is some redundancy) whose job it is to ensure an adequate and timely supply of information. In a material sense, then, all of the subtlety of our species, all of our art and science, is ultimately accounted for by a surprisingly small set of discrete genetic instructions. More surprising still, the differences between two unrelated individuals, between the man next door and Mozart, may reflect a minute difference in their genomic "recipes". We are far more alike

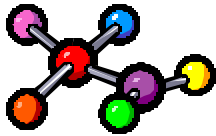
than we are different. At the same time, there is room for near-infinite variety. It is no overstatement to say that decoding our 30,000 genes is an enormous step toward unraveling the mysteries of life.

The complete set of instructions for making an organism is called a *genome*. The genome contains the blueprint for all cellular structures and activities for the lifetime of the organism. The human genome is, therefore, the full collection of genes needed to produce a functioning human being.

The Human Genome Project (HGP) is a collaborative effort of the United States Department of Energy (DOE)'s Human Genome Program and the National Institutes of Health (NIH)'s National Center for Human Genome Research. The goal of the project is to identify the function of the approximately 30,000 human genes and determine the sequence of the 3 billion DNA subunits, or bases. Research on the HGP is conducted at hundreds of university and laboratory sites throughout the United States, funded by grants from DOE and NIH. At least 18 other countries in North and South America, Europe, and Asia are doing research on the human genome, as well. The Human Genome Organization (HUGO) coordinates international efforts on human genome research.

This massive effort will provide scientists with precise information that will be key to understanding the organization and function of DNA in chromosomes. The knowledge gained and therapies developed as a result of this effort will revolutionize future biological explorations – in some ways that we can anticipate and in others that will surprise us.

## ***Genetics: The Basics***



The human genome is the full complement of genetic material in a human cell. (Despite five and a half billion variations on a theme, the differences from one genome to the next are minute; hence, we hear about *the* human genome -- as if there were only one.) The genome, in turn, is distributed among 23 sets of chromosomes, which, in each of us, have been replicated and re-replicated since the fusion of sperm and egg that marked our conception. The source of our personal uniqueness, our full genome, is therefore preserved in each of our body's several trillion cells. At a more basic level, the genome is DNA, deoxyribonucleic acid, a natural polymer built up of repeating nucleotides, each consisting of a simple sugar, a phosphate group, and one of four nitrogenous bases. In the chromosomes, two DNA strands are twisted together into an entwined spiral -- the famous double helix -- held together by weak bonds between complementary bases, adenine (A) in one strand to thymine (T) in the other, and cytosine to guanine (C-G). In the language of molecular genetics, each of these linkages constitutes a base pair. All told, if we count only one of each pair of chromosomes, the human genome comprises about three billion base pairs.

If unwound and tied together, all of the DNA in the human body would stretch more than 5 feet, but would be only 50 trillionths of an inch wide. For every organism – from the simplest bacteria to exceedingly complex human beings – the components of these slender threads encode all of the information necessary for building and maintaining life.

## **Chromosomes**



The three billion base pairs in the human genome are organized into distinct, physically separate microscopic units called chromosomes. All genes are arranged linearly along the chromosomes. The nucleus of most human cells contains two sets of chromosomes, one set from each parent. Each of these sets is comprised of 23 single chromosomes – 22 “autosomes” and an “X” or “Y” sex chromosome. (Females have a pair of X chromosomes, males an X and a Y.)

Chromosomes, when stained with dye, can be seen under a light microscope. The dye reveals a pattern of light and dark bands that indicates the variations in the amount of A and T vs. C and G. This distinctive patterning allows chromosomes to be distinguished from one another with an analysis called a "karyotype." Some chromosomal abnormalities – missing chromosomes, extra chromosomes, breaks and rejoinings called translocations – can be detected by microscopic evaluation. For example, Down syndrome, which occurs when an individual's cells contain a third copy of chromosome 21 ("trisomy 21") is diagnosed by karyotype analysis. Many other DNA changes are too subtle to be detected by this technique (i.e., to be seen on the chromosome) and require a different type of analysis. These subtle abnormalities, or "mutations," are responsible for many inherited conditions, such as cystic fibrosis and sickle cell anemia, or may predispose an individual to cancer, certain psychiatric illnesses, and other conditions.

- **Most of the information in this section is taken from To Know Ourselves, <http://www.ornl.gov/hgmis/publicat/tko/index.html>.**

## Have We Gotten Too Big For Our Genes?



The revelations of the HGP have already had, and will continue to have, a profound effect on science and medicine. Consequently, all of us will face issues of increasing complexity regarding the policies needed to govern the applications and implications of this knowledge. To this date, five percent of the budget governing the HGP has been set aside to assist in responding to the ethical, legal and social implications of this project. **The**

**advancement of the science in the HGP has surpassed the advancement of the policy used to govern this project.** Policy dialogues about the HGP have made one thing clear: the science of genetics brings forth complicated issues that affect *everyone*.

## ***Ethical, Legal, and Social Issues Related to the Human Genome Project***

The Ethical, Legal, and Social Issues (ELSI) Committee, a formal part of the HGP, has played an integral role in raising – and to some degree, addressing – policy questions related to the outcome of the scientific discoveries connected to this project. The following issues are raised as a result of the ELSI Committee's, and others', work regarding the increased availability of genetic information and its effect on humanity.

- Fairness in the use of genetic information by insurers, employers, courts, schools, adoption agencies, law enforcement agencies, the military, and others. Who will have access to genetic information, and how will it be used?
- Privacy and confidentiality of genetic information. Who owns it and controls access to it?
- Psychological impact on and potential stigmatization of individuals with genetic differences. How does this knowledge affect the individual and society's perception of the individual?
- Genetic testing of individuals for a specific condition, based upon family history and population-based screening of groups. Should genetic testing be performed when no treatment is available? Should parents have the right to test their minor children for adult-onset diseases? Are genetic test results reliable and interpretable by the medical community?
- Reproductive issues, such as use of genetic information in reproductive decision-making, and reproductive rights. Do primary care personnel understand these issues and properly counsel parents and couples about the risks and limitations of genetic testing? How reliable and useful is fetal genetic testing?
- Gene therapies to treat, cure, or prevent genetic disorders. Who decides what is normal and what is a disability or disorder? Are disabilities diseases? Do they need to be cured or prevented? Does searching for cures demean the lives of individuals presently living with disabilities?
- Genetic enhancement to supply a desirable characteristic in offspring (such

as height), which does not involve the treatment or prevention of a disorder. What ethical and safety concerns does this raise? If this became common practice, how would it affect the diversity of the human gene pool?

- Fairness in the use of genetic technology. Who will have access to these expensive technologies and therapies? Who will pay for their use?
- Education of healthcare providers, patients, and the general public. Who will provide the education to health care providers, and how will the public and patients assess competence? What is the best way to develop ongoing public and patient dialogues about genetic issues?
- Implementation of standards and quality control measures in testing procedures. How will genetic tests be evaluated and standardized for accuracy, reliability, and utility? (Currently there is very little regulation at the federal level.)
- Commercialization of products, including property rights such as patents, copyrights, and trade secrets. Accessibility of data and materials. Who owns genetic material and information?
- Philosophical quandaries regarding human responsibility, free will versus genetic determinism, and the meaning of "disease" and "health." Does someone's genes make him or her behave in a certain way? *Can* people always control their behavior? What is considered acceptable diversity in a society?
- And the list goes on...

Nationally and internationally the ethical, legal, and social implications of genetic discoveries have become highly relevant public policy discussions. At the Second International Bioethics Seminar in Japan in 1992, it was concluded that

*All human beings will be affected [by the work of the HGP], and scientists should remember that they are sequencing the shared DNA of all humanity, and they, therefore, have great responsibility, to all peoples...for this and future generations, consistent with the ... goal of health for all.*

Former President Bill Clinton, in his 1997 State of the Union address, specifically mentioned the Human Genome Project in the context of remarkable advances in medical science, when he said

*The human genome project is now decoding the genetic mysteries of life. American scientists have discovered genes linked to breast and ovarian cancer...*

In another speech that Former President Clinton made in 1997, published in *Science*, he proposed four guideposts for scientific research. Elaborating on one of the guideposts, he emphasized that

*None of our discoveries should be used to label or discriminate against any group or individual.*

In that speech Former President Clinton specifically noted that unlocking the secrets of our genetic code has the potential to identify hidden inherited tendencies that could be used by others to discriminate against or to stigmatize individuals and groups.

And finally, again at the Second International Bioethics Seminar in Japan in 1992, it was noted

*All people need to contribute their view to this [Human Genome Project] discussion. It is apparent that the ability to argue rationally about bioethics is still to be developed in ... many countries ... all of us need to think about these issues and to open our minds to the ideas of others. It is the responsibility of all people to join this discussion, because the consequences of the use of genetic information may change the expectation of health and life itself.*

The above examples illustrate that the ELSI Committee of the HGP and other national and international entities are concerned about the potential harm in the misuse of genetic information. It is time for consumer groups, professional associations, state legislators, and state government administrators to educate themselves about the accomplishments and implications of the HGP and to begin to define their roles in policy making in this arena.

The Mountain States Genetics Network Consumer Issues Committee invites you to join in the discussion and to find your seat in the policy arena. *Have We Gotten Too Big for Our Genes?* was written to help you understand how genetic research might affect each of us and how we can make a difference. Whether you are a professional, a parent, or a person affected with a genetic condition, you need to be heard!

## Creating Change Through Advocacy

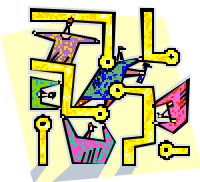


The HGP offers to provide humankind a chance, for better or worse, to control our and others' lives. Yet without advocacy to direct good public policy, we may find ourselves in no better place. The rest of this document provides tools to help you learn how to process, prioritize, and resolve possible challenges we could face as a result of the HGP.

### ***Advocacy and this Manual***

Advocacy is acting with or on behalf of a person or a class of people to promote a change in the practices, policies, and behaviors of third parties to promote a desired goal. It is a truism of advocacy that efforts should be made in the least intrusive manner, beginning with the lowest level of staff in an organization who has the authority to make the desired decision to meet a goal. If the goal is to assist an individual, then the strategies necessary to achieve the desired outcome are "individual" focused. If the goal were to assist a class or group of people, then the advocacy strategies would be "systems" focused.

### ***Multipartite Advocacy Model***



To be effective in your advocacy effort, first you must define where your efforts would be most fruitful. Dr. Elizabeth Monroe Boggs proposed the multipartite advocacy model in the 1970s. As the parent of a son with mental retardation, Dr. Boggs was active in the protection and advocacy movement that precipitated many federal laws to aid people with disabilities, such as the Developmental Disabilities Act and the Individuals with Disabilities Educational Act. The multipartite advocacy model was created to define the various roles that each person or entity

could play in protecting individuals with developmental disabilities. In *Advocacy Systems for Persons with Developmental Disabilities*, Dr. Boggs wrote:

*The protection and advocacy of individual rights...must give attention to individual concerns, complaints, and deprivations. It is not enough to generalize about the potential harm to [a] class...or about [their] rights. If there is a need for special protection for members of any minority, there is also a need to consider minorities within that minority, right down to the minority of one. At the same time, systems or class advocacy is an attractive shortcut to increasing the cost-effectiveness of limited dollars when measured by the number of people whose lives are touched...Each type of advocacy (professional, legal, lay, and protective) may be seen as having individual and collective aspects. The proposed conceptual framework recognizes and supports both.*

## **Components of the Multipartite Advocacy Model**

The multipartite advocacy model defines four advocacy prototypes. The purpose of these prototypes is to guide individuals and groups in determining where they can realize the greatest impact for their efforts.

### **Lay Advocacy**

Lay advocacy consists of informal and persuasive actions; it is often done outside of a formal system. The goal of lay advocacy seeks some type of social and/or political outcome.



- Individual lay advocacy efforts are most effective when done by members of an affected group and/or their parents, guardians, and friends. These efforts strive to effect change for a specific person. A parent working to improve the education their child is receiving in school is an example of individual lay advocacy.
- Systems lay advocacy efforts are most effective when done by entities organized to act with or on behalf of a group of people. These efforts strive to effect change for a group of people. AARP (American Association of Retired Persons) is an example of a systems lay advocacy entity.

### **Legal Advocacy**

Legal advocacy consists of formal and authoritarian actions; it is often done outside of a formal regulatory and/or service system. The goal of legal advocacy seeks some type of legislative and/or legal/class action outcome.



- Individual legal advocacy efforts are most effective when done by private attorneys, legal services, or other legal entities. These efforts strive to effect change for a specific person. A private attorney filing a lawsuit on behalf of an individual is an example of individual legal advocacy.
- Systems legal advocacy efforts are most effective when done by congress, legislatures, and legal class actions through the judicial system. These efforts strive to effect change for a group of people. The United States' Congress' creation of OSHA (Occupational Safety & Health Administration) is an example of systems legal advocacy.

### **Protective Advocacy**

Protective advocacy consists of formal and authoritarian actions; it is often done within a regulatory or service system. The goal of protective advocacy seeks standard setting, licensure, and enforcement of regulations and codes in its outcome.



- Individual protective advocacy efforts are most effective when done by entry- to mid-level bureaucrats such as a social worker. These efforts strive to effect change for a specific person. A social worker intervening on a child's behalf is an example of individual protective advocacy.
- Systems protective advocacy efforts are most effective when done by governmental agencies such as Department of Health. These efforts strive to effect change for a group of people. Department of Health staff developing and providing oversight of standards for commercial food preparation is an example of systems protective advocacy.

### **Professional Advocacy**

Professional advocacy consists of informal and persuasive actions; it is often done within a regulatory or service system. The goal of professional advocacy seeks to assure the availability of a service as its outcome.



- Individual professional advocacy efforts are most effective when done by entry- to mid-level staff, such as a nurse's aide. These efforts strive to effect change for a specific person. A home health worker questioning apparent discrepancies in a physician's medical orders for a specific client is an example of individual professional advocacy.
- Systems professional advocacy efforts are most effective when done by agency

administrators. These efforts strive to effect change for a group of people. A nursing home administrator striving to see that her facilities go beyond the standard level of care is an example of systems professional advocacy.

### ***Where Do I Fit?***

	<b>Lay Advocacy</b>	<b>Legal Advocacy</b>	<b>Protective Advocacy</b>	<b>Professional Advocacy</b>
<b>Systems</b>	Entities organized to act with or on behalf of a group of people. Goal seeks some type of social and/or political outcome for the group. **p. 20	Congress, State legislatures, and legal cases involving class actions. Goal seeks some type of legislative and/or legal class action outcome. **p. 28	Governmental agencies. Goal seeks standard setting, licensure, and enforcement of regulations and codes that affect groups of people in its outcome. **p. 33	Agency administrators in private business. Goal of professional advocacy seeks to assure the availability of a service for a group as its outcome. **p. 37
<b>Individual</b>	Parents, guardians, citizens, and friends. Goal seeks some type of social and/or political outcome targeted to individuals. **p. 24	Private attorneys, legal aid/services. Goal seeks some type of legislative and/or legal individual/class action outcome. **p. 30	Entry to mid-level bureaucrats, i.e., social workers. Goal seeks standard setting, licensure, and enforcement of regulations and codes targeted to individuals in its outcome. **p. 35	Entry to mid-level staff in a private business, i.e., nurses. Goal seeks to assure the availability of a service to an individual as its outcome. **p. 39

Different individuals may identify different goals and strategies for the same

situations; this is not unusual. Advocacy does not lend itself to a “right” or a “wrong” way. Rather, advocacy is a process that is as different as the individuals involved in defining the problem, developing the strategy, and implementing the plan.

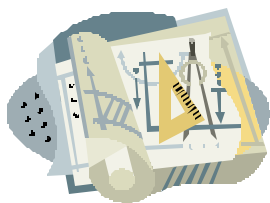
This manual outlines the tools to help you become an advocate. First, general information on advocacy is presented using the model developed by Elizabeth Boggs, Ph.D. Boggs’ model identifies four general kinds of advocacy. Factor into those four types of advocacy the option of performing either individual or systems advocacy, and eight different situations exist that generally define what kinds of advocacy Boggs’ model suggests would be most effective. Additionally, step-by-step support is given on the parts of an advocacy plan with advice on keeping the plan forward-focused.

Eight hypothetical situations are presented. These eight vignettes are intended to represent the advocacy options that generally exist according to Boggs’ model. Advocacy plans are presented in greater detail for the first of those vignettes. After the latter vignettes, we ask questions and provide a framework to help you develop your own advocacy plans. It is intended that you read all of the vignettes, not just the one(s) that you feel fit your situation. The goal of this manual is to enable you to better perform advocacy; reading all eight vignettes provides you a framework for doing so.

By studying the vignettes and their respective advocacy plans and then by developing your own plan, you will learn to be a more effective advocate. Regardless of where your personal advocacy goals fit in Boggs’ model, you will find it helpful to read this entire manual and to work through the exercises to develop appropriate advocacy plans.

## ***Advocacy Plan***

After determining the type of advocacy that best fits your situation, a methodical approach should be used to determine the problem, goal, strategies, and consequences associated with your need. Any advocacy effort not governed by such a plan can become easily sidetracked, resulting in frustration for all parties involved. Also, as a rule of thumb, remember to gauge any plan you develop by asking, “What do I win if I win and what do I lose if I win?” Bear in



mind that you may lose something, even if you win. Collaboration and compromise should be part of every advocacy strategy.

## **Steps to an Advocacy Plan**

### **Identify the problem - Problem Statement**

- a) What is the problem?
- b) Who has the problem? You, a family member, a friend, a client? Or is it a class or group of people who has the problem?
- c) What impact does this problem have on the individual or group? What injury or difficulty results from the problem?
- d) What would solve the problem? Is there only one solution, or are there several acceptable alternative solutions?
- e) Who causes the problem? Does an internal or external force generate the problem? Does an individual, or a policy, or the lack of a policy cause the problem?

### **Desired outcome or decision - Goal Statement**

- a) What outcome does the individual or class need or desire to solve the problem?
- b) What makes the desired outcome the best resolution?
- c) What agency or institution will be impacted by your desired goal?
- d) What policy, procedure, regulation, law, and/or court decision supports your proposed resolution? What policy, procedure, regulation, law, and/or court decision works against your proposed resolution?

### **Who can make the decision to implement the goal?**

- a) What is the structure of the system you intend to impact? Do you understand that system? Do you know people who work in and/or oversee that system?
- b) Who is the lowest-level person who has the authority to implement your desired goal? **REMEMBER:** Always start at the lowest level; only jump up the authority ladder (escalate) as necessary. Resist the urge to merely go to the person you know in the organization/business; Boggs' model suggests starting with the person with the lowest level of authority to implement.

## **Strategies**

- a) How are you going to reach your goal?
- b) Who is going to do what? If more than one person is involved in implementing the strategy, will everyone have a "job"? What are your timelines in regard to your "jobs"?
- c) Who is going to keep complete and detailed record of each action and timeline completion? How will this record be shared? How often will this information be shared?
- d) Who needs to be informed and advised of your actions before you implement them and as they unfold? Allies? Opponents? How much will you tell and to whom?
- e) Remember your goal. What are your acceptable compromises? How will you know if you are ready to accept a compromise?
- f) Do you know your legal rights regarding your goal and strategies? Does your goal or strategy compromise anyone else's legal rights?

## **Consequences**

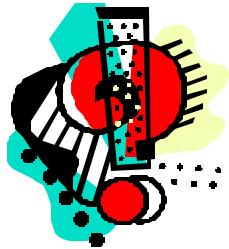
- a) Determine positive and negative effects of your actions at each step.
- b) Determine what you win if you achieve your goal, prior to implementation of strategy.
- c) Determine what you lose if you achieve your goal, prior to implementation of strategy.
- d) Determine what you lose if you do not achieve your goal, prior to implementation of strategy.
- e) Determine what you win if you do not achieve your goal, prior to implementation of strategy.

## **Determining when it is time to escalate.**

- a) Is the problem life threatening? If so, escalate quickly and jump as many levels as necessary to ensure immediate action.
- b) Determine the timelines you are comfortable with, prior to action. Make sure these timelines are reasonable and that they allow the individual or system adequate opportunity to respond.
- c) Inform all parties of your timelines at each level of escalation.

## Watch Out! Slippery Advocacy Plan Ahead

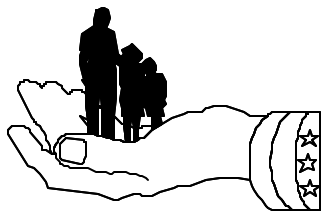
When implementing an advocacy strategy, watch for these pitfalls:



- Inability to clearly identify the focus of the advocacy effort.
- Inability to identify the strategies necessary to meet the goal.
- Fear of the possible effects of the advocacy effort, e.g., fear of public embarrassment, fear of losing, fear of being too controversial, fear of being affected personally.
- Prematurely escalating the advocacy effort without starting at the least intrusive level.
- Failure to engage in conflict when necessary to protect the person or class of persons who are being aided by the advocacy effort.
- Overemphasis on militancy or conflict in the advocacy strategy.
- Using the advocacy effort to take revenge on, punish, or disable some perceived opponent.
- Responding to pressure from outsiders to advocate for unimportant issues.
- Participating in personal attacks on individuals involved on either side in the advocacy effort.
- Accepting requests to join forces that ultimately will have no productive outcome for the advocacy effort.

### Safeguarding Your Advocacy Efforts

All persons responsible for implementation of the advocacy effort must:



- Believe that the action has purpose and the goal is the best solution.
- Be prepared to deal with the reality of “standing tall in contradiction” – that is, being informed, ready, and willing to defend the effort.
- Learn to deal with and understand the internal and external pressures that may come to bear on them, e.g., the seduction of power, money, and prestige; personal attacks on one’s integrity and character.
- Be prepared to respond; even the simplest effort may come under serious attack.
- Practice continual and habitual skepticism of any scheme, advocacy effort, or plan.

- Remember to ask and re-ask at each level of escalation what will be won if you win and what will be lost if you win.
- Be willing to modify the goal if the price of winning or losing becomes too high.

In each of the next eight sections, fictional vignettes will illustrate families, professionals, legislators, and others dealing with “real-life” situations that involve



genetic issues. These scenarios present opportunities for individual and systems lay advocacy, individual and systems legal advocacy, individual and systems protective advocacy, and individual and systems professional advocacy. Advocacy plans, in varying degrees of detail, follow the outline described above and is personalized for each situation.

**Please note:**

- **These stories take place in the near future, when the Human Genome Project has been completed, and even more information on the genetic roots of diseases and disabilities has been discovered.**
- **All of the people, organizations, and situations are fictional; any similarities to real people or situations are accidental.**

## A Systems Lay Advocacy Story and Plan

### Where Do We Belong?



**Lay advocacy consists of informal and persuasive actions; it is often done outside of a formal system. The goal of lay advocacy seeks some type of social and/or political outcome. Systems lay advocacy efforts are most effective when done by entities organized to act with or on behalf of a group of people. These efforts strive to effect change for a group of people. AARP is an example of a systems lay advocacy entity.**

Tim Welsh, Executive Director of a national multiple sclerosis advocacy agency,

stared out the window of his office, deep in thought. The waning light and building traffic signaled the end of the workday, but Tim didn't feel that he was ready to call it a day. He had received a call that morning from a Bob Stewart, seeking information and assistance with a decision whether or not to proceed with gene therapy for his young son, who had been identified to carry the recently discovered gene for multiple sclerosis.<sup>1</sup> This call had given Tim's concerns a new urgency about how his agency and its clients would cope with these new developments.

What was his agency doing to assess the immediate and long-term efficacy of gene therapy for those carrying the MS gene? What was his agency doing to ensure that genetic counselors had the most up-to-date information about gene therapy for MS? Could his agency afford to make funds available to defray the costs of this expensive intervention? Was his agency going to continue to offer its traditional programs and services for those individuals who could not or would not participate in gene therapy? These were some of the questions triggered by Bob's call to Tim.

"Just as I had begun to think that my association was proactive in meeting the needs of people with MS, researchers on the Human Genome Project discover important new information about the genetic roots of multiple sclerosis, and our association was caught unprepared," Tim lamented. He wondered if other national organizations representing consumers were facing the same onslaught of new information. "Knowledge is power – how did we get so behind these advances in research?" he wondered.

Tim decided to organize his thoughts and decide on a course of action. "People need information," he mused. "They need information about opportunities and issues resulting from the completion of the Human Genome Project. They need information about predictive testing and gene therapy. How can we identify what information people need, and where do we find the answers to their questions? Our constituents need to know what their options are, and while the science of genetics progresses, we can provide the information to help them to make a good/better/best choice."

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<sup>1</sup> This is a hypothetical situation; as of March 13, 2001, no gene for multiple sclerosis has been discovered.

Tim inventoried the resources of his organization that were available to help in this effort: its national network of affiliate offices, its board of directors, its consultants, its budget, its staff, its committees, all available to bring their unique perspectives and expertise to bear on this problem. The patient services departments and local affiliates could help to identify the issues people need help with, while their medical and scientific consultants and legislative affairs staff and consultants could help to answer those questions. Regional affiliates needed the national office to institute an accurate and timely information distribution system. Committees could recommend policies; the board could adopt those policies. The agency would rely on its scientific and medical consultants to supply answers, but would need to remember to examine policies, procedures, regulations, and laws that impact – either in a supportive or counter-supportive way – these medical and scientific decisions. Tim made a mental note not to forget that the agency's volunteers could be mustered to help in this effort.

Tim's mental checklist continued. First he would need to meet with his lead staff and the executive committee of his board to solidify a plan and an approach to this challenge and opportunity. The medical/scientific consultants would need to meet, and after reaching their conclusions, they would in turn need to meet for a strategy session with the legislative affairs staff and consultants. Once consensus was reached in the national office, this information needed to be disseminated to the local affiliates, individual members, and the general public. Budget issues would have to be resolved. Tim felt better having a plan of action.

Tim cautioned himself to remember that, "for every action there is an equal and opposite reaction." Public policy and the laws of physics had a lot in common. He resolved to stay focused on the big picture and be willing to compromise on issues of less importance to reach his greater goal. He knew that the concerns of his one agency and its constituency did not exist in a vacuum. That one did not need to win every battle in order to win the war was a lesson Tim had learned time and again in a career spent at the helm of various not-for-profit consumer organizations.

Three whirlwind weeks after his late-night planning session, Tim sat at his desk reviewing briefs and reports prepared by board members, committee members, staff, and consultants in response to his request. After reviewing these documents, Tim had reached the conclusion that the privacy, accessibility, and fiscal implications of the HGP findings were of such magnitude and scope that legislative action was required. Tim did not reach this conclusion lightly; he was

always judicious about initiating legislative activity. He decided that the next step was to schedule a conference call with the agency's Washington D.C.- based legislative consultants and make a second call to the association's director of affiliate services. He then turned his thoughts to which legislators to approach to sponsor a bill. His agency had good working relationships with a number of elected officials at various levels of government. He thought first of Rep. Joe Tannenbaum, who represented an influential constituency and had been a friend to the agency in the past on matters related to research appropriations. As the uncle of a child with a genetic disease, his personal interest was virtually guaranteed. On the Senate side, Tim gave some thought to approaching Senator Diane Napp. While a fiscal conservative, her vehement opposition to unchecked growth was noteworthy. The runaway nature of the explosion of genetics information might be similar enough to unchecked growth to capture her interest. Tim opened his address book to "N."

## ***Steps to a Systems Lay Advocacy Plan***

**How could you solve Tim's problem?**

### **Identify the problem - Problem Statement**

A national consumer support and advocacy agency for multiple sclerosis is being asked for information and policies that it does not have. Its constituency needs answers, and the national office doesn't have those answers. The implications of the Human Genome Project for multiple sclerosis were vaster than the agency was prepared to address. How can the agency accomplish what needs to be done?

### **Desired outcome or decision - Goal Statement**

The agency needs to identify the issues that have grown out of the HGP which affect its constituency, and to develop educational materials, policies, and an action plan to address those issues.

### **Who can make the decision to implement the goal?**

The agency's board of directors, staff, and consultants will be involved in identifying issues and formulating a plan. The executive director will be responsible for the refinement and implementation of the plan.

## Strategies

The board of directors, staff, and consultants are involved in defining the issues and formulating a plan of action. Reports and briefs document issues and needs; the executive director's plan details actions to meet identified goals. As the plan unfolds, the executive director will assess the need for compromise.

## Consequences

The agency will need to be diligent about monitoring the effects of its actions. "Wins" and "losses" are rarely exclusively one or the other.

## Determining when it is time to escalate

Briefs and reports document the necessity to develop both state and national legislation on privacy, accessibility, and fiscal concerns resulting from issues raised by the Human Genome Project. The agency's affiliate network will be empowered to affect this escalation at the state level, while the resources of the agency's board of directors, staff, and consultants will pool to affect the desired outcome nationally.

# An Individual Lay Advocacy Story and Plan

## Righting A Wrong?



**Lay advocacy consists of informal and persuasive actions; it is often done outside of a formal system. The goal of lay advocacy seeks some type of social and/or political outcome. Individual lay advocacy efforts are most effective when done by members of an affected group and/or their parents, guardians, and friends. These efforts strive to effect change for a specific person. A parent working to improve the education their child is receiving in school is an example of individual lay advocacy.**

Betty and Bob Stewart sit together quietly, waiting for their intake interview with a genetic counselor. Despite the bright sun streaming in through the windows, Betty and Bob feel anything but cheerful. Betty's sister has MS, and recently, during a routine pediatric exam, their second child – a cherubic, rosy-cheeked,

lively 20-month-old boy named Alex – had tested positive for an MS mutation<sup>2</sup>. During the time that they have had to wait for an appointment with a genetic counselor, Betty and Bob have struggled to grasp the implications of this information. How can they mediate the impact of MS in Alex? Would gene therapy eliminate the disease in Alex?

The obvious answer would seem to be “yes.” As parents responsible for the health and well being of their child, shouldn’t they do whatever they can to spare Alex the risk of acquiring this disease? But is there a downside, as well? Betty and Bob have asked themselves dozens of questions while trying to puzzle this out. Gene therapy is a relatively new option in health care. Have all the possible ramifications been realized? Bob, a physicist, knows that “for every action, there is a reaction.” What will the “reaction” be if they manipulate Alex’s genes to eradicate this disease? MS occurs more frequently in women than in men; what if Alex has a mild form of the disease and it never manifests in him? Research has shown that merely carrying the gene for a disease does not guarantee that you will get the disease; environmental factors appear to play a role in the development of MS. MS typically manifests in the early to mid-40s. Perhaps they should wait and let Alex make his own decision about gene therapy. Researchers may know more about the implications of gene therapy by the time their son reaches his 40s. But what if the therapy isn’t as effective if started later in life? Would waiting for Alex to have a voice in this decision compromise the effectiveness of the therapy? There is also the possibility that a cure will be found for MS by the time Alex is in his 40s.

Information available as a result of the Human Genome Project has opened up a world of choices for these concerned parents. Betty and Bob need more information in order to make an informed decision; therefore, they are meeting today with the genetic counselor. They also realized they had additional questions and concerns directly related to this counseling. How could they be assured that their counselor had the most up-to-date information? How do physicians and genetic counselors manage to keep abreast of the rapid advances in genetic research and gene therapy?

Betty and Bob also wondered about possible future discrimination that their son might face, based upon the decision they made. Could legislation, regulations, or

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<sup>2</sup>This is a hypothetical situation; as of March 13, 2001, no gene for multiple sclerosis has been discovered.

court decisions ever impact what seemed to be their private decision to make? Could laws ever be passed forcing gene therapy on citizens, so that society would not have to accommodate people with disabilities? Conversely, might laws be passed one day that affect people who had had gene therapy, if it came to be viewed by the majority as detrimental to society for some reason?

Betty and Bob recognize that, whether or not they choose to undertake gene therapy for Alex, there are implications. On the one hand, gene therapy for Alex would seem to eliminate the risk of disease from his life and to improve his overall quality of life. But the unknown implications of gene therapy continue to worry Betty and Bob. Bob, the scientist, never forgets that any one action sets in motion an entire set of other actions, with an outcome perhaps quite different from that which might have been expected or probably would have occurred had the action not been taken. Since gene therapy is relatively new, what might its long-term implications be? For instance, how might Alex's children be affected by his gene therapy today?

Betty and Bob took comfort that multiple sclerosis is not affecting Alex's health and quality of life at the moment, nor is it a life-threatening disease. They have the luxury to postpone taking any action until they feel they have all the information they need to make an informed decision.

When Betty and Bob meet with their genetic counselor, Judy Schott, they presented her with the myriad of questions they have about gene therapy for their son. As their hour with Judy draws to a close, Betty and Bob ask, "What about all the other parents in our situation? We know we are not the only ones faced with these choices, options, and decisions. Who can help all of us?"

## ***Steps to an Individual Lay Advocacy Plan***

### **How could you solve Betty and Bob's Problem?**

#### **Identify the problem - Problem Statement**

A couple's young child has been identified to carry a gene mutation for a potentially crippling disease that typically has a mid-life onset. Gene therapy is available as a result of the Human Genome Project. The child's parents have questions and concerns with the procedure, however, because the long-term implications of the therapy are unknown. The couple wants to know whether or not gene therapy is the right option for their son.

**Desired outcome or decision - Goal Statement**

The couple wants to make the right, informed decision for their child as it relates to gene therapy.

**Who can make the decision to implement the goal?**

The couple is empowered to make this decision, because of their son's age.

**Strategies**

The couple notified their immediate family about the situation they faced, and their insurance company was notified via their primary care physician's referral. Questions nagged the couple about notifying their employers, so they did not. The couple knows it is making a decision for their son and that their concerns may need to be compromised if the long-term benefit to their son seems to outweigh those concerns.

**Consequences**

Because the long-term effects and implications of gene therapy are unknown, the couple realizes that the "right" decision may not be 100% right and a "wrong" decision may not be 100% wrong, either. They realize they must gather as much accurate information as they can and make the best decision they can. The purity of "right" and "wrong" are compromised by the facts of their situation.

**Determining when it is time to escalate**

Many questions exist about the effectiveness and appropriateness of gene therapy for Alex. Such questions warrant collecting as much information as possible in order to make an informed choice. With this in mind, the couple elects to explore as many information sources as possible in addition to their pediatrician and genetics counselor. They also want to talk with other families in a similar situation, and staff at the local and national MS organization; they want to learn whether any research options are available. Because onset of the disease is not imminent, and because the disease is not life threatening, the couple feels comfortable taking all the time they need before making their decision. The couple is resolved to pursue answers until they feel they have the information they need to make the best decision on behalf of their son.

# A Systems Legal Advocacy Story and Plan

## How Much Is Private?



Legal advocacy consists of formal and authoritarian actions; it is often done outside of a formal regulatory and/or service system. The goal of legal advocacy seeks some type of legislative and/or legal class action outcome. Systems legal advocacy efforts are most effective when done by congress, legislatures, and legal class actions through the judicial system. These efforts strive to affect change for a group of people. The United States' Congress' creation of OSHA is an example of systems legal advocacy.

Senator Diane Napp, a two-term state senator, relished the opportunity her job gave her to affect real and lasting change. Senator Napp was known as a fiscal conservative and an outspoken opponent of unchecked growth. She was also considered a “people person” and was known to never ignore a constituent’s request for help. In response to just such a call for help, Senator Napp began a journey into the public policy arena for genetics.

The legislative session had just ended, and Senator Diane Napp was looking forward to just being Diane Napp, soccer mom, for a few weeks. However, a phone call from a constituent changed her plans.

Denise (Deni) Patrick explained that her brother had been diagnosed as a child with Fragile X Syndrome. Deni explained that Fragile X Syndrome is an inherited genetic condition associated with mental retardation, resulting from an alteration on the long arm of the X chromosome. Deni loved her brother. However, as she had watched her family struggle with the demands of his condition, she knew more than most the toll it could take on an entire family. While listening, Senator Napp guessed that Ms. Patrick must have been calling in response to Senator Napp’s opposition to a plan presented in the just-completed legislative session that would have increased funds available for long-term services for people with developmental disabilities. Senator Napp had opposed the legislation based upon her contention that the monies already available could be more effectively managed.

But Deni surprised her. Her call was not about that at all. Deni asked Senator Napp if she knew anything about genetic predictive testing and gene therapy. Deni had been invited to participate in a research study whose participants may carry the gene for Fragile X. The research program would ensure complete confidentiality and would provide free medical testing and other services for study participants as well as pay a stipend to defray any costs incurred as a result of participating in the study. Deni was worried about the assurance of confidentiality. How could confidentiality protect her and/or her children if she were identified as someone possibly carrying the gene for Fragile X? She was intrigued by the research program's goals.

Deni asked Senator Napp if she could help her find any existing laws regarding genetic privacy as it related to genetic predictive, and to help her to understand what protections they might offer her. Senator Napp agreed to help.

Using the Internet as a first step to researching this question, Senator Napp found several laws related to genetics. Then Senator Napp called Dr. Jayne Hiker, a high-level administrator at the State Department of Public Health, to pick her brain. Once her research was completed, Senator Napp concluded that the complete statutory protection that Deni sought in regard to the privacy of her genetic information simply didn't exist in their state, and she resolved to try to do something about this lack of protection. She called Deni to discuss her findings.

## ***Steps to a Systems Legal Advocacy Plan***

**How could you solve Deni and Senator Napp's problem?**

### **Identify the problem - Problem Statement**

Is the problem lack of information, lack of laws, lack of policies, lack of research on Fragile X, etc.? Who has the problem: Deni, her family, other Fragile X families, Senator Napp? Who or what causes the problem, and what would solve the problem?

### **Desired outcome or decision - Goal Statement**

What is the best outcome to this problem? Is another solution acceptable if not ideal? Are any agencies or institutions affected by the desired outcome? What policies, regulations or laws support or do not support the desired outcome?

### **Who can make the decision to implement the goal?**

Can Diane Napp affect the proposed change, or must the state Senate or even Congress? Will resources other than Diane Napp be needed to reach the goal?

### **Strategies**

Exactly how are you going to get done what you think needs to be? What will your role be? Diane Napp's role? A Fragile X support organization's role? The state legislature's role? Who will be the gatekeeper of information and the director of actions? Are there compromises that could or must be made to reach the goal?

### **Consequences**

What will be won and lost at each step of the process? When do the losses become greater than the gains, and what will be done as a result of this imbalance?

### **Determining when it is time to escalate**

What immediacy do you attach to this situation? What timelines are necessary, and what are the consequences if timelines are not met?

## **An Individual Legal Advocacy Story and Plan**

### **Privacy Belongs To Whom?**



**Legal advocacy consists of formal and authoritarian actions; it is often done outside of a formal regulatory and/or service system. The goal of legal advocacy seeks some type of legislative and/or legal class action outcome. Individual legal advocacy efforts are most effective when done by private attorneys, legal services, or other legal entities.**

**These efforts strive to effect change for a specific person. A private attorney filing a lawsuit on behalf of an individual is an example of individual legal advocacy.**

Damian Brown, a successful, Harvard-educated attorney practiced in the arena of medical malpractice and insurance fraud. He occasionally took *pro bono* cases referred to him by the state bar association. Today he is meeting with a potential client referred to him through that process.

Jane and Gene Solip are working class folks. Jane is a stay-at-home mom and Gene is a janitor. Jane and Gene have three children, two girls and a boy. When their son, Billy, was three, he was diagnosed with terminal cancer. Billy was diagnosed and treated at the local children's hospital. The Solips felt fortunate that Billy's doctor was a prominent and well-respected local physician, Dr. John Hartmann. Dr. Hartmann had led a research project that developed a state-of-the-art experimental treatment for children with cancer. Dr. Hartmann was also a partner in a private research company called DNA Crossroads. DNA Crossroads would sometimes absorb the cost of treatment for indigent families.

The Solips had no health insurance, so when Dr. Hartmann proposed that they consider the opportunities that DNA Crossroads could offer their son – free state-of-the-art experimental cancer treatment that might cure Billy's otherwise terminal cancer – they jumped at the chance. Dr. Hartmann did not tell the Solips that he was one of the owners of DNA Crossroads. Dr. Hartmann was eager to promote his treatment regimen that, if successful, could be patented by DNA Crossroads as a cure for certain types of cancer. The Solips would do anything to cure their son.

Billy's treatment was vigorous and initially appeared to be successful. The little boy appeared to be recovering when, suddenly, his condition deteriorated and it was necessary to put him on life support. Dr. Hartmann was out of town when the crisis occurred, so another team of doctors treated Billy. During this crisis, the Solips were asked detailed questions about Billy's treatment, and there were some subtle indications that some of the physicians treating Billy had misgivings about the course of his treatment. After a week's struggle, the little boy died; the cause of death was cancer. A few weeks later the Solips met with Dr. Hartmann to discuss some of the criticisms they had heard regarding Billy's treatment. Dr. Hartmann seemed genuinely anguished about their son's death. He told them that experimental therapies are always risky, and Billy's cancer had been terminal. He said the grumbling they had heard was, no doubt, a result of the frustration the doctors had felt over being unable to save him.

A year later the Solips read in the paper that Dr. Hartmann's company, DNA Crossroads, was applying for a patent for a gene therapy treatment for cancer. The Solips' initial reaction to the article was sadness that this hadn't come in time to save their son. Dr. Hartmann was quoted as saying he was excited to be part of the cure for this dreaded disease. He was in the process of initiating a

cooperative treatment agreement with numerous cancer centers around the country. The article went on to say that of 200 patients who had received the experimental treatment, only one had died due to a "treatment anomaly." The Solips wondered if this individual could be their son and, if so, what exactly was meant by a "treatment anomaly." They remembered the muttered misgivings about Billy's treatment that they'd heard during that final crisis. They began to wonder: Had their son been treated appropriately, or had he been a victim of medical malpractice? They called the state bar association the next day for advice and had been referred to Damian Brown.

## ***Steps to An Individual Legal Advocacy Plan***

**How could you solve Jane and Gene's problem?**

### **Identify the Problem - Problem Statement**

What is the problem and who has it? Is the problem Damian Brown's, Jane and Gene Solip's, or Dr. Hartmann's? While it may be that ultimately all of these people have a problem, within the parameters of, in this case, legal advocacy, who has the problem? What causes the problem, and would eradicating that source be a solution? Does the problem affect an individual only or does it have a broader implication?

### **Desired outcome or decision - Goal Statement**

What is needed for resolution to the problem? While Billy had a terminal disease, was his quality of life or death different/earlier because of the drug trial? Are there good, better, best solutions? What makes a resolution good, better or best? Who outside of the immediate family is impacted by the resolution? What supports your decision about the best resolution, a policy, regulation, law, etc.?

### **Who can make the decision to implement the goal?**

What is the structure of the system you're going to impact? Will you affect DNA Crossroads only? Is your first contact the lowest-level person within that company? How does Dr. Hartmann fit into your plan?

### **Strategies**

How will you reach the goal you've identified? Is there a role for the Solips, Damian Brown, and Dr. Hartmann? Have you identified timelines for the work to be accomplished? Who will keep records of who is doing what with whom when? Have

you identified that good, better, best spectrum so you can consider compromises to your identified goal? What are your legal rights regarding your goal, and does your goal compromise anyone else's legal rights? Specifically, do your legal rights compromise Dr. Hartmann's or DNA Crossroads'?

### **Consequences**

Remember that for everything we "win", we may also "lose." As you look and work toward your goal, are the costs greater than the benefits so that you need to change your strategy and/or goal?

### **Determining when it is time to escalate**

The problem is no longer life threatening; Billy's life is lost. Immediacy, then, is less an issue. Rather, what timelines will you set for actions to be complete prior to escalating? Are those reasonable timelines and does everyone know them? Does everyone know of your intent to escalate if timelines are not met?

## **A Systems Protective Advocacy Story and Plan**

### **How Do We Make The Rules Fair?**



**Protective advocacy consists of formal and authoritarian actions; it is often done within a regulatory or service system. The goal of protective advocacy seeks standard setting, licensure, and enforcement of regulations and codes in its outcome. Systems protective advocacy efforts are most effective when**

**done by governmental agencies such as Department of Health. These efforts strive to effect change for a group of people. Department of Health staff developing and providing oversight of standards for commercial food preparation is an example of systems protective advocacy.**

Dr. Jayne Hiker was a twenty-year veteran in the State Department of Public Health. She had survived several changes in executive administration and had a reputation as an effective, no-nonsense administrator.

Dr. Hiker had been closely following the discoveries of the Human Genome Project and was concerned about the public policy implications of protecting individuals in

this new era. She had been excited and pleased by the passage of the Genetic Privacy Act. Her advice to Senator Napp had been instrumental in the drafting of the legislation. Additionally, she had testified before both the state's house and senate committees, advocating for passage of the bill. The passage of the legislation was going to allow for regulated standards for genetic research, and Dr. Hiker had been looking forward to developing those standards.

A recent development was troubling, however. A lawsuit had been filed regarding the privacy of research subjects and profit-driven research. The lawsuit troubled Dr. Hiker because she hadn't been aware that this type of research had been going on in her state. When the lawsuit was filed, the press had a field day; suddenly medical research was the suspected culprit in a myriad of medical ills.

Under ordinary circumstances, the development of the rules and regulations for the new legislation would be done without public scrutiny. Staff would draft the regulations that would, in turn, be reviewed by experts participating on an advisory committee. Seldom did Dr. Hiker need to deal with public input or concern. However, this lawsuit was going to draw press attention to the scope and effectiveness of rules and regulations as a tool to protect the medical privacy of citizens.

Dr. Hiker took her responsibility as a public servant seriously; she had a duty to make sure that the rules and regulations for the Genetic Privacy Act met the letter of the law while also satisfying the public's need to be reassured that their medical privacy was not being compromised for profit.

## ***Steps to a Systems Protective Advocacy Plan***

**How could you solve Dr. Hiker's problem?**

### **Identify the problem - Problem Statement**

What is the problem, and whose problem is it? Have you adequately assessed the problem, its impact, its solution(s), and its causes?

### **Desired outcome or decision - Goal Statement**

Is your goal one that is needed to solve the problem? What makes your solution the best one? Have you accurately assessed the obstacles and supports to

reaching your goal?

### **Who can make the decision to implement the goal?**

Have you adequately and accurately assessed the structure of the system you will be impacting? With whom will you start implementing your goal?

### **Strategies**

What is your plan to meet your goal? Who's doing what, including record keeping and follow-up?

### **Consequences**

What are the effects of actions and reactions?

### **Determining when it is time to escalate**

Where and when will you go if your intended results are not realized?

## **An Individual Protective Advocacy Story and Plan**

### **Ready, Set, Go?**



Protective advocacy consists of formal and authoritarian actions; it is often done within a regulatory or service system. The goal of protective advocacy seeks standard setting, licensure, and enforcement of regulations and codes in its outcome. Individual protective advocacy efforts are most effective when done by entry- to mid-level bureaucrats such as a social worker.

These efforts strive to effect change for a specific person. A social worker intervening on a child's behalf is an example of individual protective advocacy.

Dan Thomas was a new hire in the State Department of Public Health. Dan had a Master's degree in Health Administration, and his title was Administrator of Health Regulations. His job included the oversight of public and private medical

research programs. Dan had worked in private research before completing his Master's degree, and he knew many of the players in the local private research community. In his first weeks on the job, Dan spent hours studying the newly passed rules and regulations on genetic privacy; he had closely followed the passage of the Genetic Privacy Act. One of his first assignments was to survey all public and private research projects in the state to assure their compliance with the new standards, and with the standards set by the Institutional Review Boards and the national Office for Human Research Protections.

Dan was in the process of planning this survey when he received a call from Dr. Claude Alpert, a former colleague. Claude was a well-respected physician in private practice and was also on the faculty of the university's school of medicine. Claude congratulated Dan on his new job and offered to buy him lunch to celebrate; Dan found himself wondering if Claude had something on his mind other than a celebratory lunch. The conversation at lunch was light and casual, until Claude suddenly asked Dan what he was going to do with the genetic privacy standards. Dan asked Claude what he meant. Claude looked troubled, and asked Dan to keep what he was going to tell him confidential. One of Claude's sons had been diagnosed with HIV two years earlier; conventional therapies had been ineffective. His son's health was deteriorating. An experimental gene therapy study that his son would qualify for was being proposed at the university, but because of the pending surveys and compliance issues with the new genetic privacy rules and regulations, the university had put a moratorium on new research projects until they assured the compliance of existing projects. Claude had supported the Genetic Privacy Act and thought the rules and regulations were good, but he thought the implementation process was taking too long, too long for his son and others like him. Dan was sympathetic and said he would do what he could to facilitate this process. He asked Claude what kind of timeline he envisioned for the resolution of this issue. Claude replied only, "Soon, please make it soon."

## ***Steps to an Individual Protective Advocacy Plan***

**How could you solve Dan's problem?**

**Identify the problem - Problem Statement**

What and whose is the problem?

**Desired outcome or decision - Goal Statement**

What are your priorities?

**Who can make the decision to implement the goal?**

Are you dealing with the right person or people to affect the change you want?

**Strategies**

How will you accomplish your goal?

**Consequences**

What are they?

**Determining when it is time to escalate**

When and to where is escalation appropriate?

## **A Systems Professional Advocacy Story and Plan**

**Go? Where?**

Professional advocacy consists of informal and persuasive actions; it is often



done within a regulatory or service system. The goal of professional advocacy seeks to assure the availability of a service as its outcome. Systems professional advocacy efforts are most effective when done by agency administrators. These efforts strive to effect change for a group of people. A nursing home administrator striving to see

that her facilities go beyond the standard level of care is an example of systems professional advocacy.

Lisa Messinger was the current chairperson of a national professional association for genetic counselors. She had kept abreast of the discoveries of the Human Genome Project, and she made every effort to be aware of developments in genetics that affected her organization's many members, both nationally and locally. She was up-to-date on her own state's recently passed Genetic Privacy Act and its ramifications.

This morning Lisa had received a call from Dan Thomas, the Administrator of

Health Regulations at the State Department of Public Health. He wanted to know what she might know about a self-imposed moratorium on in-state health research programs as a result of the pending compliance assessments in the wake of the state legislature's imposition of genetic privacy rules and regulations. Dan said that he had a personal acquaintance that had made him aware of this situation. He wondered if Lisa would allow him to use her organization's listserv to both provide information to and solicit information from her association's in-state members.

He also wondered if her organization would like to co-sponsor a conference on recent advances in gene therapy as a result of the HGP. Her first impulse was to decline; she thought this would use up too much of her national organization's resources on a local matter. Then she happened to see the note on her desk from Judy Schott asking if she were aware of any longitudinal studies on the implications of gene therapy. It occurred to her that if Judy, a past president of this very organization, didn't know the answers to these questions, then few others in their organization did either. Perhaps the training Dan was suggesting could be a good use of resources if it could become a model for the association's state affiliate offices to use. They certainly had to do something to keep their membership up-to-date on these rapidly developing scientific and technological advances as well as support their mission of encouraging public dialogue about genetic issues.

She told Dan that their Information Services Committee would be meeting next week. Would he be willing to attend that meeting and help to develop a pilot project that their two agencies could co-sponsor to meet the need for timely and accurate information on genetic advances?

## ***Steps to a Systems Professional Advocacy Plan***

**How could you solve Lisa's problem?**

**Identify the problem - Problem Statement**

**Desired outcome or decision - Goal Statement**

**Who can make the decision to implement the goal?**

## Strategies

## Consequences

## Determining when it is time to escalate

# An Individual Professional Advocacy Story and Plan

## What is the Right Answer?



**Professional advocacy consists of informal and persuasive actions; it is often done within a regulatory or service system. The goal of professional advocacy seeks to assure the availability of a service as its outcome. Individual professional advocacy efforts are most effective when done by entry- to mid-level staff, such as a nurse's aide. These efforts strive to effect change for a specific person. A home health worker questioning apparent discrepancies in a physician's medical orders for a specific client is an example of individual professional advocacy.**

Judy Schott, a genetic counselor, had a short break between appointments. She sat, head in hands, momentarily overwhelmed by the demands of her profession. Ten years ago when she started out, her practice consisted in large part of pregnant women having amniocentesis to check for Down syndrome. The explosion of new genetics' information as a result of the HGP, gene therapy for instance, had significantly increased the variability of the types of cases she now saw. And with the science being so new, it was impossible to know the long-term implications of options such as gene therapy.

The technological and scientific advances seemed to be reducing the ability of society to manage the implications of these new developments. The combination of this new knowledge and technology, and few policies, rules, or regulations to protect consumers creates a situation ripe for abuse. And Judy did not see enough dialogue to these issues on the part of her colleagues, the medical community in

general, and especially the public such as elected officials and consumers.

Judy was also concerned that, despite her best efforts to stay up-to-date in her field, she did not have the resources to know everything about the newest technologies, therapies, and hypotheses. Scientific and technological advances were having a very real effect on the daily lives and everyday decisions of ordinary people – like her clients and herself. She wondered how her colleagues and professionals in the other fields of medicine and even insurance companies were coping with this situation.

Judy saw that it was time for her next appointment. She was seeing a couple, the Stewarts, whose son had been found to test positive on a MS mutation. They were trying to make a decision about whether or not to proceed with gene therapy for their son. Judy had posted a request on the genetic counseling listserv for information and learned about the existence of the MS Support Organization as well as a clinical trial starting in a few months, but she had been unable to get specifics. “Focus,” she told herself. “My immediate responsibility is to these clients. I will tell them honestly what I know and also what I don’t know, and do my very best to help them understand the process of this difficult and complicated decision.”

## ***Steps to an Individual Professional Advocacy Plan***

**How could you solve Judy’s problem?**

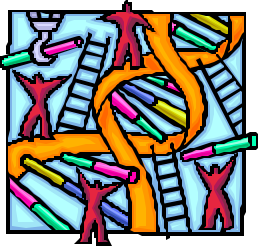
OK. This one is yours! What do you need to do to develop an advocacy plan? What are the steps and checks you need to keep in mind to assure that you are on the best course to resolve the problem herein presented? Good luck!

## Supplemental Information

Guidelines for Genetics Services Delivery is a brochure created and produced by the Mountain States Genetics Network to fill the growing need to educate the public on an issue that is becoming a large part of their medical world in the 21<sup>st</sup> century. In an effort to aid the education of Advocates, we have included the Guidelines with the Advocacy Manual because it is an integral part of the issues raised in Advocacy. To further your understanding of Genetics issues we have also included the Genetics Glossary used in the medical genetics and scientific community.



# Guidelines for Genetics Services Delivery



As a result of research in medical genetics, there will be dramatic changes in the practice of medicine, in how we think about health and disease, and in the number of people who need genetic services. Genetics clinics are already experiencing the increased patient load. In order to maintain quality services, it is important to define a quality program. This document spells out guidelines for the many dimensions of genetic services - from the population served to the necessary system requirements.

Genetics services should always include clear and complete communications with the family regarding all findings and results of any evaluations. This may include any or all of the following:

- individual or family sessions with the geneticist or genetic counselor, repeated as appropriate,
- copies of evaluation and/or study results, and
- a letter summarizing what was said during the counseling session.

Genetics services should be sensitive to the language, culture, and beliefs of the families seen in the clinics. When necessary, medical translators should be present. Written information should be presented in terms that are understandable to the family and translated into the family's primary language if it is not English. As with all medical care, families must be assured that the results of all laboratory tests and any information they provide will be kept confidential. As appropriate, family members or guardians should be involved in decisions regarding care and treatment.

Written reports should also be sent to the referring physician and/or agency.

## ***A. Population Served***

Genetics services should be available to those who are concerned that they or their children have, or may develop, a potentially hereditary condition. More specifically, services should be available to people who:

- Have a medical, psychological, or other condition that is genetic such as hemophilia, sickle cell anemia, or phenylketonuria,
- have family members who have similar conditions,
- have a child with a birth defect, learning disability, medical disorder, or other conditions that may have a genetic basis,

- are expecting a child and have been exposed to a teratogen (such as alcohol, tobacco, infections, medications, chemicals, or radiation) that could harm the child,
- have had two or more stillbirths, or miscarriages,
- are infertile,
- are expecting a child and are at increased risk for problems because the mother is over age 35 or has a chronic health condition (such as diabetes), or are at risk for developing a serious medical condition that may 'run in the family' such as heart disease, high blood pressure, mental illness, cancer, or diabetes.

Thus, people who seek genetic services do not necessarily have a genetic condition nor have to be 'sick'. However, they or their children may be at risk for developing a medical disorder.

### ***B. Availability Of Services and Accessibility***

Genetics services should be community-based. In each community there should be health care workers who have been trained to screen for genetic disorders and complete preliminary examinations. If the community is too small to support such a person, regional clinics that are staffed by certified personnel should be offered on a regular basis.

When necessary, families should receive assistance with transportation arrangements to a genetics center. At the minimum, a family should be offered assistance in using public transportation or in taking advantage of special services that are available within the community. When travel distances are great, teleconferencing should be an option if a face-to-face meeting is not required.

### ***C. Clinical Requirements***

While most medical visits focus on a specific medical concern of a person, a genetics consultation is often much more encompassing and time-consuming in that it may include any or all of the following:

#### **Medical history / background information**

- a review of all appropriate medical records, psychological evaluations, laboratory reports, and radiographs (such as x-ray and ultrasound films),
- a family history and a pedigree (a family tree that is used for medical purposes),
- a prenatal history that includes possible exposures to substances that may

harm a fetus and a description of any problems during the pregnancy, as well as standard information such as length of the pregnancy, maternal weight gain, fetal position and activity,

- postnatal history (a description of how the mother and infant did immediately after delivery and during the first few months of life),
- a history of growth and the attainment of developmental milestones,
- a review of school performance,
- a history of behavioral problems, and
- other medical history regarding hospitalizations, surgeries, and major illnesses.

### **Laboratory tests and physical exam**

- a complete physical examination including a head-to-toe exam for features that are not symmetrical or are variations of normal, and
- laboratory studies as appropriate.

### **Counseling and education**

- a diagnosis (or a discussion of several possible diagnoses) and a treatment plan,
- a discussion of the possible types of therapy, long-term significance, and/or risks to other family members and/or to future pregnancies,
- referral to other specialists who can help manage particular disorders,
- a discussion of concerns the individual/family might have regarding financing for medical services, anxiety around a diagnosis, or other coping issues,
- assistance in making decisions in a non-directive fashion,
- referral to appropriate support services and family support groups, and on-going follow up.

### **Specific Situations**

While the above requirements apply to all genetics evaluation sessions, some services are more appropriate under specific situations. These are described below.

### **Pregnancies**

Genetics services involving pregnancies may also require:

#### ***Counseling***

- explaining benefits, limitations, and risks of prenatal diagnostic tests,
- discussing pregnancy options,

- identifying and discussing exposures to environmental agents (such as radiation, medications, infections, and chemicals) that may harm the fetus, and
- arranging for delivery at a medical center equipped to deal with potentially serious medical problems.

### ***Screening and diagnostic procedures***

- drawing blood from the mother to screen for specific conditions (such as defects of the spinal column or conditions that occur more frequently in particular ethnic groups),
- doing an ultrasound to look for specific problems, and/or performing prenatal diagnostic tests (amniocentesis, CVS).

The parent(s) has (have) the right to decide how to follow-up on the results of any evaluation; this includes the right to carry a fetus with a genetic disorder to term or to terminate the pregnancy.

### **Newborns**

Genetics services involving a newborn or a very young child may also require:

- screening the newborn for a genetic condition such as PKU (phenylketonuria),
- complete autopsy or laboratory analysis of tissues from a placenta, miscarried fetus, or stillborn infant, and/or
- evaluation to detect problems involving the brain and/or others parts of the nervous system.

### **Young Children**

Genetics services involving a child may also require:

- referral for a developmental evaluation, and/or
- management of health conditions that are age specific.

### **Adolescents and Adults**

Genetics services involving adolescents and adults often require:

- a discussion of reproductive risks,
- referral for assistance with medical, psychological, vocational, or social problems,
- management of health conditions that are age specific,
- screening to determine if a person is a carrier of a genetic disorder,
- presymptomatic testing to determine whether or not a person is at-risk for developing a particular condition (in general, testing is more specific than

- screening), and/or
- counseling to help cope with the disorder or the risk of developing a disorder.

#### ***D. Follow-up Requirements***

As a result of genetics services, the family may be referred elsewhere for management of the genetic condition. For example, the family may need access to:

- primary medical care in a health clinic or with a private physician,
- specialty care as needed for treatment,
- psychological and/or supportive counseling,
- various therapies to assure optimal level of function, and on-going health education.

Agencies and/or programs that can facilitate the provision of services (initial as well as follow-up) include the following:

- a newborn screening program
- a birth defects registry
- Early Intervention Programs (Public Law XX-XXX, part C of IDEA)
- Title V for Children with Special Health Care Needs
- EPSDT (Early Periodic Screening, Diagnosis, and Treatment) for persons who are Medicaid eligible
- schools with personnel who can assist children who have special needs
- child development clinics
- public health agencies
- support and advocacy groups
- adoption agencies (for providing health information about the birth family to authorized individuals)
- primary health care providers and
- other programs that may be specific to a particular state or region

#### ***E. Genetics Services Providers / Personnel Requirements***

Clinics and facilities that provide genetics services should have a physician and counselor who are certified by, or have Active Candidate Status with, the American Board of Medical Genetics and/or the American Board of Genetic Counseling. In addition, they should have formal working relationships with:

- other physicians, genetic counselors, and health care professionals,
- subspecialists in medical genetics, clinical cytogenetics, teratology, biochemical genetics, cancer genetics, and molecular genetics, registered

nurses, medical social workers, registered dieticians, and other health professionals, and

- laboratories that participate in nationally approved quality assurance programs.

## ***F. Nongeneticist Health Care Providers***

In order for any genetics services delivery system to work, it is essential to have the participation of nongeneticist health care providers who:

- make appropriate referrals,
- provide follow-up and treatment as part of primary care or in a specialty clinic,
- maintain communication with the geneticists to assure appropriate management of the genetic condition, and
- assist the family in understanding the genetic nature of the condition.

## ***G. The System Requirements***

In addition to the requirements discussed above, other elements must be in place in order to provide families and individuals (including children) with quality genetics services. These include:

- a genetics network that includes teaching facilities, accredited laboratories, health care providers working in a variety of settings (in private practice, associated with academic medical centers, and with health care groups, not-for-profit/ for-profit),
- a computerized system that can be used for making a diagnosis, coordinating patient care, finding resources, tracking patterns of occurrence, etc.
- a system that protects individual choices regarding reproductive options,
- procedures that do not allow discrimination on the basis of a genetic condition,
- legislation that enables health care providers to administer genetic programs such as newborn screening,
- ways to identify individuals and families who could benefit from genetics services including birth defect registries, community clinics, and health care providers (such as private obstetricians, pediatricians, family practitioners, and public health nurses),
- access to a library system that can provide general as well as technical information to families and health professionals,
- a tracking system to locate individuals who require follow-up services and/or who are at risk for genetic disorders (repeat genetics evaluations may be

recommended as new information or questions arise; a registry assists in identifying families who need to be contacted),

- an education system that will provide appropriate information about genetics to people at all levels - professional (undergraduate, graduate and continuing education) patient/family, and general population and public schools, and
- advisory groups that include individuals with genetic disorders, parents who have children with genetic conditions, and health professionals.

## ***H. Confidentiality and Ethical Issues***

The information that can be learned from genetic testing and family histories is unique in that it can affect family dynamics and has broad implications for future generations. Therefore, genetics services must protect an individual's privacy and autonomy. The utmost care must be taken to protect against the use of genetic information by unanticipated third parties and the possible consequences of such occurrence must be discussed with the families. <sup>1</sup>

## ***I. Cost Coverage / Funding***

Genetics services should be available to everyone, regardless of income. These services, including those provided by certified counselors, should be covered by all health care, third-party payers including:

- private insurance,
- state and federally funded health insurance/plans,
- managed care and other pre-paid health insurance programs
- self-funded insurance plans. <sup>2</sup>

When necessary, services should be available on a sliding fee scale.

## **In Conclusion**

Genetics is an ever-changing science. New genetics findings addressing existing and new diagnoses occur daily. The impact of these increasing numbers of studies on health care providers and families is overwhelming. Genetics impacts the health and lives of all individuals in different ways at different times. Genetic information must be provided with the utmost care and sensitivity to the families served by health care providers. This guide has been developed to assist families in understanding the essential elements of genetic services. For more information regarding genetics services in your area, visit the Mountain States Genetics Network website at [www.mostgene.org](http://www.mostgene.org).

## REFERENCES

*(These two references are from information provided by Lynda M. Fox, J.D)*

1. Language that is in accord with the CORN (Council of Regional Networks) Code of Ethical Principles for Genetics Professionals.

"Genetics impacts the health and lives of all individuals in different ways at different times. Genetic information is unique in its familial dynamics and in its implications for future generations. Genetics services must, therefore, be provided with the highest degree of respect for the principles of autonomy and privacy, and must be conveyed with sensitivity in language that the recipients can genuinely understand. The utmost care must be exercised in protecting against the use of genetic information by unanticipated third parties, and the potential consequences of the use of genetic information by various parties must be discussed with the recipients of genetics services. Among the issues requiring particular consideration by the providers and recipients of genetics services are:

- the testing of children for genetic mutations associated with untreatable late-life onset disorders;
- the possibility of the use of genetic testing information by employers and insurance providers; (Many states have addressed this concern through state statutes limiting how employers and insurance providers may use genetic testing information. Each genetics services provider must be familiar with the applicable statutes for his/her particular state, and the limitations of those statutes. In particular, providers and recipients of genetics services must understand that self-funded insurance plans are not subject to state law and thus not protected by state statutes. Federal legislation also offers certain protections against the use of genetic information by employers and insurance providers. Under the Americans With Disabilities Act, employers may not use genetic information as the basis of hiring decisions if the potential employee is otherwise qualified to perform the essential functions of the job. The recently passed Health Insurance Portability and Accountability Act provides certain protections against genetic information being considered as a pre-existing condition in the absence of a clinical diagnosis, and against the exclusion from group coverage of an individual because of genetic information. These provisions went into effect June 10, 1997.)
- the difficulty of ensuring the privacy of genetic information in this age of computerized medical records;
- the difficulties that may arise within families when certain family members seek genetic information that may be relevant to other family members who do not wish to know that particular genetic information.

Only with the sensitive consideration and genuine understanding of these issues will genetics services be effectively provided, and decisions regarding genetics services be made in an informed and voluntary manner."

2. Self-funded insurance plans. These plans are private as opposed to state and federally funded plans. They are group insurance plans, usually through an employer, in which the entity sponsoring the plan holds the premiums paid for coverage in a pool and pays claims out of that same pool. Compare this with plans through insurance companies in which premiums are paid to the insurance companies and claims are paid accordingly. Self-funded plans are not under the jurisdiction of state law, rather they are governed by federal ERISA provision. Insurance companies are under state law jurisdiction.

### **Contributors to "Guidelines for Genetics Services"**

This guide is intended to provide information to help individuals recognize what constitutes sound genetic services. These services are available to varying degrees for the management and diagnosis of genetic disorders. This information has been written and organized by members of the Mountain States Genetics Network (MoStGeNe) and applies to the six-state region of its operation: Arizona, Colorado, Montana, New Mexico, Utah, and Wyoming. MoStGeNe members include representatives from state health departments, genetics service laboratories, hospitals, private practices, educators, university medical schools, and consumers.

Established in 1984, MoStGeNe supports the following goals: a) to promote communication among genetic services providers, b) to share resources and problems, c) to coordinate services across state lines, d) to explore and develop collaborative studies and services such as common data bases and regional technical and educational assistance, e) to improve the quality and quantity of genetic services in the region.

The Mountain States Genetics Network wishes to thank Judy Capra for her contributions to this document.

The Mountain States Genetics Network is indebted to Lynda Fox, JD for her contributions to the development of these guidelines.

# Glossary of Genetic Terms

**Abnormal:** A variation from the normal. In the case of genes an abnormal gene may result in a specific disorder.

**Acquired mutations:** Gene changes that arise within individual cells and accumulate throughout a person's lifetime; also called somatic mutations.

**Age of onset:** Age at which the effects of a genotype become evident.

**Adenine (A):** A base; one of the molecular components of DNA and RNA.

**Advanced maternal age:** Women over age 34 (age 35 at delivery) are at increased risk for nondisjunction trisomy in the fetus.

**AFP (Alpha-fetoprotein):** A protein excreted by the fetus into the amniotic fluid and from there into the mother's bloodstream through the placenta.

**Alleles:** Any one of two or more alternate forms of a gene located at the same locus.

**Allelic (gene) frequency:** The percentage of alleles (genes) in a population that are of a particular type.

**Amino acids:** Small molecules that form the building blocks of proteins.

**Amniocentesis:** Prenatal diagnosis method using cells in the amniotic fluid to determine the number and kind of chromosomes of the fetus and, when indicated, perform biochemical studies.

**Amplification:** Production of multiple copies of a sequence of DNA.

**Anencephaly:** One of a series of defects in which the tube of tissue destined to form the brain and spinal cord fails to close, in this case resulting in absence of brain, skull cap, and scalp.

**Aneuploidy:** State of having a variant chromosome number (too many or too few). (i.e., Down syndrome, Turner syndrome).

**Anomaly:** Deviation from the average; anything unusual, irregular or contrary to the general rule.

**Anticipation:** Onset of an inherited disorder at an earlier age, or worsening of symptoms in successive generations.

**Artificial insemination:** The placement of sperm into a female reproductive tract or the mixing of male and female gametes by other than natural means.

**Autosome:** Any chromosome that is not part of the pair of sex chromosomes. Humans have twenty-two pairs of autosomes, numbered from 1 to 22.

**Autosomal Dominant Trait:** Refers to how a disease, condition, or trait is inherited. (Autosomal) the gene involved with the disease is located on a chromosome other than X or Y (sex chromosomes). Therefore the gene will be located on chromosome 1 to 22. (Dominant) all genes occur in pairs. In autosomal dominant conditions, if one of the genes in a pair is not working the disease will be present, it does not matter that the other gene in the pair is functional. Males and females are equally affected. If an individual has a nonfunctional gene, each of his/her children has a 50% chance of inheriting it. Examples of dominant diseases include Huntingtons disease, neurofibromatosis, and most inherited cancers..

**Autosomal Recessive Trait:** Refers to how a disease, condition, or trait is inherited. (Autosomal) the gene involved with the disease is located on a chromosome other than the X or Y (sex chromosomes). Therefore the gene will be located on chromosome 1 to 22. (Recessive) all genes occur in pairs. In autosomal recessive condition, if one of the genes in a pair is not working, the functional gene is enough to prevent the disease. The condition only occurs when an individual inherits two nonfunctional genes. Men and women are equally affected. If a couple carry the same nonfunctional gene, for each pregnancy, they have a 25% chance of having an affected child, a 50% chance to have a child who is carrier, and a 25% chance to have a child who is not affected and not a carrier. Examples of recessive diseases include cystic fibrosis and sickle cell disease.

**Autosome:** All chromosomes except those involved in sex determination. Human cells have 22 pairs of autosomes.

**Balanced Translocation:** Occurs when two chromosomes break and exchange places, leaving the same amount of genetic material. An individual with a balanced translocation will be unaffected, but their children may be affected in a variety of ways.

**Bases:** The molecular building blocks of DNA and RNA: adenine, cytosine, guanine, thymine, and (in RNA only) uracil. In DNA, A attaches only to T, and C attaches only to G. In RNA, A attaches only to U, and C attaches only to G. Two strands of DNA are held together in the shape of a double helix by weak chemical bonds between their base pairs.

**Base pair:** Two of the building blocks of DNA held together by weak bonds. In a DNA molecule, adenine always bonds with thymine (A-T), and cytosine always bonds with guanine (C-G).

**Base sequence:** The order of bases in a DNA molecule.

**BRCA1:** A gene that normally helps to restrain cell growth.

**BRCA1 breast cancer susceptibility gene:** A mutated version of BRCA1, which predisposes a person towards developing breast cancer.

**Carrier:** An individual who carries one copy of a gene mutation for a specific disorder. The carrier is also referred to as a heterozygote. Carriers do not typically develop the disorder, but can pass the gene mutation on to their children. However, only two carrier parents could typically have a child affected with the disorder (see autosomal recessive above).

**Carrier Test:** A genetic test usually from a blood sample, performed to determine if a healthy individual carries a nonfunctional gene.

**Cell:** The basic unit of life. All living organisms are a collection of cells.

**Centromere:** The constricted portion of the chromosome, separating it into its two arms. The region of a chromosome to which spindle traction fibers attach during mitosis and meiosis; where chromatids are held together.

**Chorionic villus sampling:** An invasive prenatal diagnostic procedure involving removal of villi from the human chorion to obtain chromosomes and cell products for diagnosis of disorders in the human embryo.

**Chromatid:** The two halves into which a chromosome is longitudinally divided at mitosis and meiosis; one of two strands of DNA held together at the centromere, which exists after a chromosome has made a replica of itself.

**Chromatin:** The complex of nucleic acids (DNA and RNA) and proteins (histone and nonhistone) of which chromosomes are composed.

**Chromosome:** A structure of DNA and protein found in the cell nucleus, which carries genes. Normally, each body cell has a total of 46 chromosomes-- 22 paired autosomes and one pair of sex chromosomes (XX=female and XY=male).

**Chromosome banding:** A technique for staining chromosomes so that bands appear in a unique pattern particular to the chromosome.

**Clinodactyly:** In-curving of a finger, resulting from angulation at the interphalangeal joints.

**Clone:** A group of identical genes, cells or organisms derived from a single ancestor. Identical twins are clones, as are colonies of bacteria that reproduce by simple cell division.

**Cloning:** The process of making genetically identical copies.

**Colonoscopy:** Examination of the colon through a flexible, lighted instrument called a colonoscope.

**Congenital:** Present at birth, not necessarily inherited.

**Consanguinity:** Relationship by descent from at least one common ancestor.

**Contiguous genes:** Genes physically close on a chromosome that when acting together express a phenotype.

**Crossovers:** The exchange of genetic material between two paired chromosomes during meiosis.

**CYS (Chorionic Villus Sampling):** This is a diagnostic procedure sometimes used early in pregnancy (at 9 to 11 weeks of growth) to detect certain genetic disorders.

**Cytogenetics:** A branch of genetics concerned with the study of chromosomes.

**Daughter cells:** The two cells which result from division of a single cell.

**Deletion:** The loss of a segment of the genetic material from a chromosome, which is variable in size and may be detectable at the DNA level (small deletion) or at the chromosomal level (large deletion).

**De novo translocation:** A chromosomal translocation that appears in an offspring but was not present in either parent.

**Derivative chromosome:** Chromosome of origin for an abnormal or misplaced piece of chromosomal material.

**Differentiation:** The complex of changes involved in the progressive diversification in the structure and functioning of the cells during fetal development.

**Diploid:** Having a full set of genetic material consisting of paired chromosomes that contain one chromosome from each parent.

**Direct gene test:** A test that can detect specific mutations or alterations in the DNA of a gene.

**Disruption:** A single event interrupting normal development of an organ leading to structural anomalies. Such disruptions may be of vascular, infectious or even mechanical origin.

**Dizygotic:** Two cells having been fertilized resulting in twins (fraternal).

**DNA (deoxyribonucleic acid):** The long, spiraling molecule that orchestrates the cell's daily operations and provides

the genetic blueprint for the physical characteristics of all living organisms. Its two strands intertwine like a spiral staircase to form a structure called a double helix. Sub-units, called bases, are the rungs of the staircase and carry the genetic code. (See also *RNA*).

**DNA fingerprinting:** A process that uses fragments of DNA to identify the unique genetic makeup of an individual.

**DNA repair genes:** Certain genes that are part of a DNA repair pathway; when altered, they permit mutations to pile up throughout the DNA.

**DNA sequencing:** Determining the exact order of the base pairs in a segment of DNA.

**Dominant gene (or dominant allele):** A gene which, when present, produces a certain trait, and “dominates” over a recessive allele in the gene pair.

**Double helix:** A common name for DNA; it refers to the molecule's double-stranded spiral staircase structure.

**Duplication:** Occurs when a chromosome or part of a chromosome is duplicated. This may happen during cell division.

**Dysmorphic feature:** Developmental abnormality of the form and structure of a certain feature in an individual (often minor structural abnormalities).

**Embryo:** An organism in the early stages of development (in humans, the developing organism from conception until approximately the end of the second month).

**Endonuclease:** Restriction enzyme.

**Enzyme protein:** A type of protein whose function is to act as a catalyst and make chemical reactions possible in living organisms. In the absence of the enzyme, the chemical reaction for which the enzyme is responsible will not take place.

**Etiology:** Causation; the study of causes, especially of disease.

**ELSI:** Ethical, legal and social implications (of the Human Genome Project).

**Ethics:** The study of fundamental principles which defines values and determines moral duty and obligation.

**Eugenics:** The “improvement” of humanity by altering its genetic composition by encouraging breeding of those presumed to have desirable genes.

**Expansion:** Addition of multiple copies of trinucleotide repeat sequences during meiosis.

**Expressivity:** The extent to which a gene's characteristic is seen in an individual. If a gene is said to have variable expressivity, then the trait may vary from very mild to severe but is never completely unexpressed.

**Familial:** Characteristic of some or all members of a family.

**Familial adenomatous polyposis:** An inherited condition in which hundreds of potentially cancerous polyps develop in the colon and rectum.

**Familial cancer:** Cancer, or a predisposition toward cancer, that runs in families.

**Fetoscopy:** Method of pre-natal diagnosis where the inside of womb and the developing fetus can be examined visually and sampling can be performed using a fetoscope.

**FISH (fluorescence in situ hybridization):** A laboratory technique that uses fluorescent tags to identify specific chromosomes or portions of chromosomes.

**Fragile sites:** A small break or constriction of a chromosome; a non-staining gap of variable width that usually involves both chromatids and is always at exactly the same point on a specific chromosome derived from an individual or kindred.

**Functional gene tests:** Biochemical assays for a specific protein, which indicates that a specific gene is not merely present but active.

**Gamete:** A male or female reproductive cell. In the female, an ovum (or egg); in the male, a sperm.

**Gene:** A defined section of DNA along the chromosome that encodes information for the production of a protein necessary for the functioning of the organism.

**Gene amplification:** Any process by which specific DNA sequences are replicated disproportionately greater than their representation in the parent molecules; during development, some genes become amplified in specific tissues.

**Gene cloning:** Isolating a gene and making many copies of it by inserting it into cells and allowing it to multiply.

**Gene deletion:** The total loss or absence of a gene.

**Gene expression:** The process by which a gene's code directs the manufacture of proteins that determine an organism's characteristics (see *expressivity*).

**Gene mapping:** Determining the relative positions of genes on a DNA molecule.

**Gene markers:** Landmarks for a target gene, either detectable traits that are inherited along with the gene, or distinctive segments of DNA.

**Gene pair:** The two genes, one derived from each parent, with information for producing a protein. One gene comes from the chromosome set contributed by the egg cell; the other gene from the chromosome set contributed by the sperm cell. All genes come in pairs with the exception of genes on the X chromosome in males. Males have only one X chromosome; therefore the genes on the X chromosome in males are present only in a single dose.

**Gene therapy:** A newly evolving technique designed to treat inherited genetic disorders. The medical procedure involves substituting a healthy gene for an altered gene in the cells of a patient's body.

**Gene testing:** Examining a sample of blood or other body fluid or tissue for biochemical, chromosomal, or genetic markers that indicate the presence or absence of genetic disease.

**Genetic counseling:** Education and guidance offered by professional advisors in order to help people make informed decisions based on genetic knowledge. Genetic counseling is intended to help a person understand the meaning of specific information about his or her genes. It also is intended to help a person decide whether to have a genetic test performed or what to do with information provided by such a test.

**Genetic engineering:** The technology used to genetically manipulate living cells to produce new chemicals or perform new functions.

**Genetic linkage map:** A chromosome map showing the relative positions of the known genes on the chromosomes of a given species.

**Genetic screening:** The process of identifying individuals in a population who may be at increased risk for having or being a carrier of a genetic disorder.

**Genetic testing:** The process of determining whether a person or organism has a certain gene.

**Genetics:** The scientific study of heredity; how particular traits or qualities are transmitted from parents to offspring.

**Genome:** All the genetic material in the chromosomes of a particular organism. The human genome consists of three billion bases, organized in about 100,000 genes on a set of chromosomes.

**Genotype:** The actual genes carried by an individual (see also *phenotype*).

**Germ cells (or germline):** Cell line from which gametes are produced.

**Germline mutations:** (See *Hereditary mutation*.)

**Guanine (G):** A base; one of the molecular components of DNA and RNA that forms the genetic code. Always bonds with cytosine (G-C).

**Haploid:** A single, complete set of chromosomes (half the full set of genetic material), present in the egg and sperm cells of all animals. Human beings have 23 chromosomes in their sex cells. (Compare to *diploid*).

**Hereditary mutation:** A gene change in the body's reproductive cells (egg or sperm) that becomes incorporated in the DNA of every cell in the body; also germline mutations.

**Heterogeneity:** The production of identical or similar phenotypes by different genetic mechanisms.

**Heterozygote:** Having two different alleles at a given locus (adjective: heterozygous)

**Homologous chromosomes:** A "matched pair" of chromosomes, one from each parent, having the same gene loci in the same order.

**Homozygote:** Having identical rather than different alleles at a given locus. Both alleles have the same mutation. (adjective: homozygous)

**Human Genome Project (HGP):** A worldwide project aimed at deciphering all the three billion bases of the human genome, including mapping and sequencing every gene. This information will help to more rapidly identify genes causing diseases in humans.

**Huntington's disease:** An adult-onset disease characterized by progressive mental and physical deterioration; it is caused by an inherited dominant gene mutation.

**Implantation:** The attachment of the fertilized ovum to the endometrium or uterine wall, and its embedding in the compact layer of the uterus. This usually occurs at six or seven days after fertilization.

**Imprinting:** Refers to the phenomenon whereby the effect of inheriting a gene is different depending on whether the inheritance was from the mother or father, e.g., Prader Willi syndrome and Angelman syndrome.

**Inborn errors of metabolism:** Conditions in which the metabolism of an organism is abnormal because of the presence of one, or a pair of abnormal alleles.

**Incomplete penetrance:** The gene for a condition is present, but not obviously expressed in all individuals in a family with the gene.

**Inversion:** Occurs where a chromosome breaks in two and becomes reattached after turning around 180 degrees. Providing no chromosome material is lost, this abnormality should have no effect on the individual. However, that individual may carry the risk of producing eggs or sperm with the incorrect amount of chromosome material.

**In vitro:** In the test tube or laboratory.

**In vivo:** Inside a living organism.

**Karyotype:** A photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type.

**Late-onset disorder:** A disorder, which is not apparent at birth but develops later in the course of an individual's life.

**Leukemia:** Cancer that begins in developing blood cells in the bone marrow.

**Li-Fraumeni syndrome:** A family predisposition to multiple cancers, caused by a mutation in the p53 tumor-suppressor gene.

**Linkage analysis of pedigree:** The tracking of a gene through a family by following the inheritance of a (closely associated) gene or trait and a DNA marker.

**Linkage test:** An indirect form of genetic testing in which a known region of DNA located near a gene for a disorder can be used as a "marker" or indicator for that gene. This type of testing is used when the target gene has not yet been identified or when a direct test is not practical because a specific mutation is not known.

**Locus:** The position that a gene occupies on a chromosome.

**Mapping:** (See *gene mapping, physical map*).

**Marker:** An identifiable physical location on a chromosome that can be identified and followed as it is inherited. This can be a nearby gene which produces a known protein, a restriction enzyme break point, or a series of repeated bases.

**Meiosis:** The production of sex cells, which are not genetically identical, through a series of cell divisions. Compare to mitosis.

**Melanoma:** A cancer that begins in skin cells called melanocytes and spreads to internal organs.

**Metabolic disorder:** A disorder caused typically by an enzyme abnormality in the cells. Usually associated with slower development and growth.

**Meiosis:** The process of cell division which results in a cell where the 23 pairs of chromosomes split into two, each with 23 chromosomes. These cells are the female ova (eggs) and the male sperm.

**Mitochondrial inheritance:** The mitochondria are cellular organelles containing DNA, which encode genes that may sustain disease causing mutations. Since only maternal mitochondria are inherited, mitochondrial diseases exhibit non-Mendelian inheritance patterns.

**Mitosis:** Division of all cells except the reproductive cells with chromosome numbers and genetic make-up identical to that of the parent cell.

**Monosomy:** The total loss of one of a pair of chromosomes. This occurs, for example, in Turner Syndrome where one X chromosome is lost, leaving a total of 45 chromosomes.

**Mosaicism:** Where two or more cell populations occur within the body cells, each with a different genetic make-up.

**Multifactorial disorder:** A disorder brought on by the joint action of multiple factors. The contributing factors include several different genes as well as various types of agents from the environment.

**Mutation:** A spontaneous or induced change in the DNA of a cell. A gene which has undergone change from the normal gene. This mutation may be an established one or a new sporadic mutation.

**Nondisjunction:** Failure of two members of a chromosome pair to separate during cell division so that both are passed on to the same daughter cell.

**NTD (Neural tube defect):** Occurs when there is defective development of the spinal cord and the brain in utero. Conditions such as spina bifida and anencephaly are examples of such developmental defects.

**Nucleotide:** A molecular subunit of DNA or RNA consisting of a base (adenine, guanine, thymine, or cytosine in DNA; adenine, guanine, uracil, or cytosine in RNA). Thousands of nucleotides are linked to form a DNA or RNA molecule. (See *DNA, base pair, RNA*).

**Nucleus:** The portion in the center of each cell which contains the chromosomes with their genetic material.

**Oncogenes:** Genes that normally play a role in the growth of cells but, when overexpressed or mutated, can foster the growth of cancer.

**Ovum:** Female egg: reproductive cell carrying 23 chromosomes.

**PCR (polymerase chain reaction):** A laboratory technique that permits a small DNA section located between two

fixed points on the DNA molecule to be duplicated many times, yielding many copies of that DNA section.

**Penetrance:** An individual who carries a dominant gene may show a variable degree of the symptoms of the disorder.

**Phenotype:** The observable expression of a gene. Example—brown eye color.

**Photomicrography:** The technique of making photographs through a light microscope.

**Physical map:** A map of the locations of identifiable landmarks on DNA, such as genes, or restriction enzyme cutting sites.

**Placenta:** A complex structure occurring in pregnancy. It is attached to the wall of the womb and connected to the fetus by the umbilical cord. The fetus receives its nourishment through the placenta and the vessels of the umbilical cord.

**Precancerous polyps:** Growths in the colon that often become cancerous.

**Predictive gene tests:** Tests to identify gene abnormalities that may make a person susceptible to certain diseases or disorders.

**Predisposition (genetic):** The individual concerned is intrinsically more prone to develop a particular disorder.

**Presymptomatic test:** A genetic test performed to determine if a gene (or genes) is present which will bring on a health problem late in an individual's life.

**Probability:** The odds or chance that an event will happen.

**Proband:** Individual in a family who brought the family to medical attention.

**Probe:** A specific sequence of single-stranded DNA, typically labeled with a radioactive atom, which is designed to bind to, and thereby single out, a particular segment of DNA.

**Prognosis:** Prediction of the course and probable outcome of a disease.

**Prophylactic surgery:** Surgery to remove tissue that is in danger of becoming cancerous, before cancer has the chance to develop. Surgery to remove the breasts of women at high risk of developing breast cancer is known as prophylactic mastectomy.

**Proteins:** The active molecules in all cells. Proteins control biochemical reactions and determine the physical structure of organisms.

**Renal cell cancer:** A type of kidney cancer.

**Retinoblastoma:** An eye cancer caused by the loss of a pair of tumor-suppressor genes; the inherited form typically appears in childhood, since one gene is missing from the time of birth.

**Ring formation:** Occurs where the ends of a chromosome bend over and fuse together with a loss of genetic material.

**Recessive gene (or recessive allele):** A gene, which must be present on both chromosomes in a pair to show outward signs of a certain characteristic.

**Recombinant DNA:** A form of DNA produced by splicing together segments of DNA from two or more organisms.

**Restriction enzyme (endonuclease):** A protein that breaks the chromosomes apart into many small fragments by cutting the DNA of the chromosomes wherever a specific short sequence of bases occurs. (See also *Restriction enzyme cutting site*).

**Restriction enzyme cutting site:** A specific sequence of DNA at which a particular restriction enzyme cuts the DNA.

**RFLP (Restriction fragment length polymorphism):** Variations occurring within a species in the length of DNA fragments generated by a species endonuclease.

**RNA (Ribonucleic acid):** A chemical cousin of DNA, RNA is responsible for translating the genetic code of DNA into proteins.

**Ribosome:** The small cellular structure in which RNA translates the genetic code into proteins.

**Robertsonian translocation:** Occurs when translocations of chromosomes involve end-to-end fusion with the loss of the short arms. The balanced carrier has 45 chromosomes and is normal; any children may be affected in number of ways.

**Screening:** Looking for evidence of a particular disease such as cancer in persons with no symptoms of disease.

**Selection:** The process of determining the relative share allotted individuals of different genotypes in the propagation of a population; the selective effect of a gene can be defined by the probability that carriers of the gene will reproduce.

**Sequencing:** Determining of the order of bases in a DNA or RNA molecule.

**Sex chromosomes:** The X and Y-chromosomes in human beings that determine the sex of an individual. Females have two X chromosomes; males have one X and one Y.

**Sex determination:** The mechanism in a given species by which sex is determined; in many species sex is determined at fertilization by the nature of the sperm that fertilizes the egg.

**Single-gene disorder:** A disorder which comes about when there is a mutation in a specific gene, and one (for a dominant disorder) or both (for a recessive disorder) of the genes in the gene pair cannot function properly.

**Single strand:** One half of a DNA double helix.

**Somatic cells:** Any cell in the body except sex cells.

**Somatic mutation:** A mutation which occurs in any of the body cells of an individual over the course of that person's life. Since the mutation is not in the eggs or the sperm cells, it cannot be passed on to children. (See also *acquired mutations*).

**Sonography:** Pictures made using echoes from a very high frequency sound.

**Sperm:** (Abbreviation of **spermatozoon**) the male reproductive cell carrying 23 chromosomes.

**Spina bifida:** Open spine, one form of major central nervous system defect.

**Susceptibility test:** A genetic test for a gene whose presence can increase the chances of developing a health problem later in life. The problem may not develop even if the damaged gene is present, and it may occur even if the gene is absent.

**Syndrome:** A recognizable pattern or group of multiple signs, symptoms or malformations that characterize a particular condition; syndromes are thought to arise from a common origin and result from more than one developmental error during fetal growth.

**Teratogens:** Any agent that raises the incidence of congenital malformations.

**Thymine (T):** A base; one of the molecular components of DNA and RNA. Always bonds with adenine (T-A).

**Trait:** Any detectable phenotypic property of an organism.

**Transcription:** The process by which DNA passes genetic information to RNA. Transcription is the first step in producing proteins.

**Translation:** The process by which RNA makes proteins.

**Translocation:** The transfer of all or one part of a chromosome to another location on the same chromosome or to a different chromosome after chromosome breakage.

**Trinucleotide repeats:** Long chains of three bases, e.g., CAG,CAG,CAG,...and so forth in DNA.

**Trisomy:** A condition in which there are three, rather than two copies of any one chromosome in the same cell.

**Tumor-suppressor genes:** Genes that normally restrain cell growth but, when missing or inactivated by mutation, allow cells to grow uncontrolled.

**Uracil (U):** A base; one of the molecular components of RNA. Bonds only with adenine (U-A).

**Virus:** A noncellular biological entity that can reproduce only within a host.

**Wild type:** Normal allele or normal phenotype.

**X-linked dominant:** A pattern of inheritance attributed to genes located on the X chromosome. A disorder will appear when one copy of the nonfunctional gene for that disorder is present. Affected males pass X-linked dominant genes to all their daughters but none of their sons. Affected females pass X-linked dominant genes, on average, to half of their daughters and half of their sons.

**X-linked inheritance:** This refers to any gene found on the X chromosome or traits determined by such genes; it also refers to a specific mode of inheritance of such genes.

**X-linked recessive:** A pattern of inheritance attributed to genes located on the X chromosome. Males with the gene will be affected because all the genes on their single X chromosome will be expressed. Females who have two X chromosomes can be carriers. Affected males in a family are related through females.

**Zygote:** A fertilized egg.

## References

*Blazing a Genetic Trail*. Online Book, Howard Hughes Medical Institutes  
See: [www.hhmi.org/GeneticTrail/glossary/glossary.htm](http://www.hhmi.org/GeneticTrail/glossary/glossary.htm)

*Diving Into The Gene Pool – Glossary*. The Exploratorium, San Francisco, CA  
See: [www.exploratorium.edu/genepool/glossary.html](http://www.exploratorium.edu/genepool/glossary.html)

"Genetics: A Family Affair." A symposium held in Schaumburg, Illinois, March 9-14, 1997. Sponsored by the Illinois Department of Public Health Genetics Section and Triton College Continuing Education Center for Health Professionals.

*Genetics Glossary*. CaF (Contact a Family with rare conditions), London, England  
See: [www.cafamily.org.uk/genegloss.html](http://www.cafamily.org.uk/genegloss.html)

*Glossary of Genetic Terms*. Genetics Education Center, Univ. Kansas Medical Center  
See: [www.kumc.edu/gec/glossary.html](http://www.kumc.edu/gec/glossary.html)

Jackson, John F., M.D. *Genetics and You*. New Jersey: Humana Press, 1996.

Ricker, Wendy. *Alphabet Soup*. New England Regional Genetics Group: Consumer Concerns Committee, 1997.

*Understanding Gene Testing*. National Institute of Health  
See: [www.accessexcellence.org//AE/AEPC?NIH?gene27.html](http://www.accessexcellence.org//AE/AEPC?NIH?gene27.html)

Weiss, Joan O., and Machta Jayne S. *Starting and Sustaining Genetic Support Groups*. Baltimore: Johns Hopkins University Press, 1996.

*Your Genes, Your Choices*. Publication of Science + Literacy for Health, a project of the AAAS Directorate for Education and Human Resources.  
See: <http://her.aaas.org/ehr/books/about.html>

Zallen, Doris Teichler. *Does It Run in the Family? A Consumer's Guide to DNA Testing for Genetic Disorders*. New Jersey: Rutgers University Press, 1997.

