GENETIC DRIFT
Mountain States Regional Genetic Services Network

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Consumer Issues in Genetics

From the Editor and Authors

Continuing advances in medical genetics are resulting in new standards of care for patients of every age and socioeconomic group. Yet, despite these advances, issues regarding universal access to services, privacy and reimbursement continue to reflect a medical system in flux. Both providers and consumers are struggling to address these concerns, even as new genetic technologies are daily changing the face of medicine. This issue of the *Genetic Drift* offers medical professionals insight into the consumer perspective regarding the issues surrounding provision of genetic services. Often dependent upon support from private and public health programs, consumers are routinely challenged to obtain adequate access to the diagnostic and counseling services which constitute optimum care in the context of modern medicine. Moreover, the cost of services, which may not be covered by third-party payers, adds significantly to the dilemma faced by patients searching for genetic services with limited financial resources. Therefore, the following articles also provide the medical professional and the consumer with insight into the many questions surrounding fiscal responsibility for special-needs patients, the emotional and legal impact of genetic services on the patient and their family, the social responsibility of providers to equalize access, and the realities of cost and reimbursement.

Our thanks to the guest contributors for sharing their experience and perspectives. References to additional information and resources, including website referrals, are listed at the conclusion of the issue.

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Susan M. Bryan, Editor

In This Issue

New Website Address

Please note that the Mountain States Genetics Network website has a new address: http://www.mostgene.org/

The Implications of Genetics Testing

It's More Than A Simple Blood Test

Contributed by Vickie Venne, M.S.

Editor's Note: For medical professionals, the routine of diagnosis is less straightforward for cases in which genetic testing is required. The ramifications of the test results necessitate additional education and counsel for the patient and their family in order to fully prepare those involved, and thereby avoid unnecessary confusion, fear and stress. The following contribution by a genetic counselor who deals daily with patients encountering genetic testing for the first time highlights the complexity of issues surrounding what initially might be viewed as "a simple blood test."

Virtually half of the articles about genetic testing in the lay press contain the words "simple blood test." In a high-tech medical world of complicated imaging, medications and surgeries, reporters seem intrigued that much genetic testing can be performed on a blood specimen. And for doctors and patients, ordering a blood draw or sticking your arm out for a few tubes of blood seems easy.

So why the fuss? Because after the easy blood draw, the laboratory analysis is often complex and expensive, the results can sometimes be difficult to interpret, and the results usually impact more than an individual patient. Regardless of the reason for the blood draw, primary care physicians are the conduit for the information, and should know the implications of the test and counsel accordingly.

The variety of circumstances for which genetic testing is appropriate are numerous. Situations as diverse as a chromosome analysis for a baby with multiple malformations, a newborn's PKU screen, a teen with growth abnormalities and a suspected metabolic problem, and an adult with a family history of colon cancer wanting presymptomatic testing generate a wide spectrum of issues that need to be reviewed prior to ordering the blood test. In addition to the standard disclaimer on consent forms regarding the impact of a blood draw (slight pain, possible bruise, fear of needles), many genetic consent forms contain language about psychological impact, family relationship issues and the social risks of discrimination. These issues vary with the circumstances, but are important to consider for each type of genetic test.

Who is the testing candidate?

Many, but not all, genetic tests are ordered for diagnostic purposes as a consequence of clinical findings. In those situations, the person for whom the testing is most useful is the affected one. However, when ordering a test for presymptomatic purposes, the person on whom relevant information will be obtained may not be your patient. In families in which there is a significant amount of cancer, the most appropriate testing candidate may be a parent or a cousin who was diagnosed at a young age. For example, a negative colon cancer DNA result on a young man may not be as useful if he is not in a family in which the specific gene causing the increased colon cancer risk is known. Defining a testing strategy and then being willing to advocate for testing, especially if the best candidate lives in a different community or has a different insurance plan, can be time consuming and vital to obtaining valuable information for your patient.

The impact of making a diagnosis.

Not confirming a diagnosis can be frustrating for a family, who may be wanting a better understanding of the etiology, prognosis and potential options for an affected child. Conversely, confirming a diagnosis provides a "label" with valuable information about natural history and treatment options, but can also create potentially negative social stigmatization. Appreciating the impact that either a positive or negative result will have on a patient or family is a key aspect of providing comprehensive care.

Genetic ramifications.

In addition, test results will often have genetic (and sometimes medical) ramifications for other family members. Parents may be identified as obligate carriers of an autosomal recessive gene, siblings may be at risk for developing a particular condition, a parent may be the source of an inherited dominant mutation, extended family members may now be at risk of inheriting mutations. In addition, a new medico-legal twist of informing these extended family members who may be at risk as a result of the genetic finding (so-called "duty to warn") is reshaping the physician-patient healthcare partnership in the genetics community. So what began as an individual blood test can result in information with significant medical implications for not only that person, but for many family members.

The family dynamic.

The psychological dynamics in each family are different, but it is important to remember that genetic testing on an individual is performed in the context of a family unit. The results of many blood tests create an emotional response - genetic testing is not alone in that. However, the unique emotional aspect of many genetic tests is that they are wrapped up in an already-existing family dynamic. Issues such as the expectations for a child (which may be different for the mother and the father) which change based on a test result, survivor guilt, the role of caretaker siblings, and anxiety about developing a condition if the testing was performed presymptomatically, can be added to the current manner in which a family handles its interactions. Although many family coping strategies are developed long before genetic testing is offered, the results may trigger emotional or communication issues that will need additional attention. If the primary-care provider does not have in-house resources to address the family dynamics, referral to a genetic center or other mental healthcare provider may be warranted.

So the next time you are preparing to order a simple blood test for a genetic condition, pause to consider the following checklist:

- Is the patient an appropriate testing candidate?
- Would you or a staff member be able to be an advocacy resource?

- Is the patient (and when appropriate, the family) prepared for either a positive or a negative test result?
- Does the patient understand the medical, psychological and social ramifications?
- Do you have a referral list of appropriate specialists and/or genetic counseling resources to resolve any issues that cannot be handled in the office?

If you can answer yes to these questions, the blood test (although not simple) can be ordered with confidence. ❑

Difficulties in the Diagnosis of Genetics Disorders

Contributed by Ginger Dodge

Editor's Note: The following experience of this patient and her child underscores the limitations of current genetic diagnostics. As many practitioners already know, the results of genetic testing do not always provide a clear diagnosis or indicate a specific course of therapy. The frustration and stress borne by patients presenting with multiple indications of a genetic disorder require that professionals employ not only medical expertise in determining the nature of the disorder, but also significant support and empathy in helping the patient cope physically and emotionally with the day-to-day challenges of the condition -regardless of its origin.

I am a 34-year-old wife and mother. My son, Nick, was born on October 31, 1994 by normal vaginal delivery - although induced because of IGR (inter-uterine growth restriction). Nicholas weighed 3 pounds, 10.5 ounces. He was in NICU from the time he was born and finally got to come home from the NICU the week of Thanksgiving (on oxygen)

When I first found out I was pregnant, I felt something was wrong with our baby. But everything seemed normal, and after a few months those feelings went away. I figured it was just overactive hormones! Finally, at eight and a half months, I convinced my doctors that there really was something wrong. I was very small for being so far along; some people at work didn't even know I was pregnant. The baby wasn't moving much anymore either. More ultrasounds were done at the local hospital. It was obvious something was wrong, but nobody would tell us anything. Later that evening, my doctor called me and ordered me down to the University of Colorado Hospital in Denver immediately! Many ultrasounds were done and I spent hours lying there, staring at monitors and trying to see if our baby was okay. They also said my amniotic fluid was low and that I had a two-vessel umbilical cord instead of the normal three-vessel cord. I was in the hospital for two weeks before labor was induced and Nick was born.

Although irregularities in Nick's physiology and development were detected by ultrasound, and were immediately apparent at birth, a clear diagnosis was not forthcoming.

During his first year, Nick had two hernia and undescended testicle operations; he was hospitalized for pneumonia when he was two. In the past four years, I've lost count of the number of doctors and specialists we have seen as we searched for a diagnosis of Nick's condition: at least seven in person, two or three others by mail.

The rigors of trying to determine the cause of Nick's disorder so that he could be treated appropriately, and the stress of the diagnostic runaround, were hard on our family. Most stressful was when we were told Nick was Trisomy 18 (this was done without verifying the diagnosis with a blood test). Nick's life expectancy at that time was two weeks. We were told a few days after that diagnosis that it was not correct. He was seen by a geneticist at birth, and about every three months thereafter. He was nine months old before a diagnosis of Cornelia de Lange syndrome (CdLS) was made. It was confirmed in 1996, but in 1998 was "un-confirmed." We are working with Dr. Jackson Laird in Colorado on getting a firm diagnosis. At least we have been able to rule some syndromes out. It's also been comforting to confirm that Nick is generally healthy (no heart problems, etc.), though developmentally delayed.

The general outcome of our experience has been useful, if trying. The most difficult period was the "roller coaster" of our emotions when we were wrongly told Nick only had two weeks to live. Happily, he is now a healthy, happy child, although we have a few concerns with skeletal development, craniosynostosis and swallowing. Although we have no developmental prognosis, his life expectancy is optimistic.

For medical professionals and other families facing issues regarding genetics and the diagnosis of unknown disorders, our experience may offer some insight. When Nick was first born, and we knew there was SOMETHING genetic going on, I expected the doctors to look at blood test results and tell me what my son's diagnosis was exactly. Patients need to be aware that the doctors may NOT have a diagnosis, or it may take a while to find one. Parents and family members involved in the situation need to know that there is so much that is still unknown about genetics, and that a diagnosis may not be very easy. ❑

Clergy Response to Genetics Issues in Counseling

Pastoral Genetic Testing

Contributed by Peter Abdella, M.S.

Editor's Note: The increasing complexity of medical genetics issues requires patients to turn to professionals in many areas to gather enough information to make informed healthcare decisions. Increasingly, genetic counselors are required to give patients insight into not only the physiological implications of issues in genetics, but also the psychological, sociological and emotional factors, which must be taken into account when patients are challenged by a genetics-related medical issue. The following commentary reflects the experience of a genetics counselor who is also a member of the professional clergy.

What consolation can you offer a childless couple in their early forties whose longed-for child, at 35 weeks of gestation, has been diagnosed with an extra copy of chromosome 18 (too much genetic material) that causes severe mental retardation, multiple physical (especially heart and respiratory) problems, and death 90 to 95 percent of the time before the child reaches the age of one? Suppose they come to you wanting help deciding how much care to plan on providing for their child given this condition-assuming she or he doesn't die in the womb. Could you advise them?

Now change the scenario a bit. What if it were a couple in their mid-thirties whose first pregnancy was very difficult, but successful: they have a beautiful, 20-month-old baby girl. But now, at 18 weeks of gestation, their second pregnancy goes awry. Prenatal diagnosis reveals that the developing baby has a open neural tube defect and hydrocephalus (fluid build-up in the brain) so serious that the baby — if it makes it to birth — will almost certainly be faced with paraplegia, loss of bladder and bowel control, and a less than 10 percent chance of even borderline reasonable intellectual functioning, not to mention a host of elaborate surgeries. How would you counsel this couple should they tell you they are seriously considering terminating this pregnancy, due to the exhausting nature of the care of this baby would require and its likely toll on the family structure? Suppose they do so; would you be able still to celebrate the loss of this very-much-wanted child with some form of liturgy?

Almost weekly reports in the news media of genetic advances can leave us with the impression that cures for genetic disorders are virtually at hand. I do not want to dismiss the import of the Human Genome Project or of the various gene therapy trials that are currently taking place (and receiving intense scrutiny because of a recent protocol-related death). But the reality of the current practice of clinical medical genetics is far different. What genetic testing and diagnosis offers today is still predominantly information. Families and individuals must then struggle with the burdens and benefits that information poses to the well being of their offspring and themselves.

Cases like the ones above have been part and parcel of my work in La Jolla for the past year or more. Most of the couples that I see are referred by genetic counselors in San Diego and Orange counties. These referrals have been enormously provocative and representative of the range of complex issues raised by modern genetics. ❑

Providing for Extended Care

The Montana Self-Sufficiency Trust

Contributed by Jan Duffy

Editor's Note: As the practice of medicine becomes more sophisticated, some patients once diagnosed with severe, chronic genetic disorders with shortened life expectations are now able to live extended, even normal, life spans. The economic issues of long-term care for individuals with disabilities become critical when the individual, his or her family, the medical community and public and private funding sources are faced with the costs of long-term care. The following article discusses options, as they exist in Montana and many other states, for developing economic support strategies for children with chronic disabilities who may survive their family caregivers.

Parents who wish to leave a legacy to their child with a disability are faced with a dilemma: if the child has assets in his or her name, these assets may make the individual ineligible for government benefits like Supplemental Security Income (SSI) and Medicaid. Very few families have enough money to set up a trust to cover all of their child's needs, so they are reluctant to create even a small trust, which will not be enough to sustain the individual but will have the effect of disallowing the person from more substantial and long-term government benefits.

Montana's Self-Sufficiency Trust (MSST) is a mechanism to help families deal with the dilemma of providing for a disabled loved one's future. MSST was established by state statute to allow families to create trusts for their children and use the interest income from the trusts to purchase supplemental services for the beneficiaries without jeopardizing their eligibility for government benefits.

"Government benefits" means eligibility for means-tested programs like SSI and Medicaid, which pay for basic needs such as shelter, food, clothing and medical care for individuals with disabilities. MSST funds are designated for "extras," above and beyond the costs of basic sustenance, so MSST funds are considered "supplemental" to the basics paid for by SSI. MSST has a letter of agreement from the federal government saying that individuals with MSST accounts can remain eligible for SSI benefits. The reason that MSST beneficiaries remain eligible for benefits is because the interest income from MSST passes from the private to the public sector; in this process the funds become, in effect, government monies. Since services are purchased for the beneficiaries without any monies being attributed to them individually, they are considered to be receiving supplemental services and remain eligible for government benefits.

To participate in the Self-Sufficiency Trust, donors (parents or other interested individuals) set up individual trust accounts for their children with disabilities. These individual trust accounts are pooled in a statewide Private Trust. In addition to the Private Trust to which donors contribute, there is

also a Charitable Trust which receives charitable donations from individuals and organizations. The combined Private and Charitable Trusts make up the total Self-Sufficiency Trust. The principal in the pooled trusts of MSST is invested and the income earned is transferred to the State Trust Fund. The state then uses the income from the SST to purchase supplemental services for the trust beneficiaries from non-profit providers in the community.

The State Trust Fund is a special account in the Montana state treasury. The state auditor has the power to direct payments from the trust account to the Developmental Disabilities Program (DDP) and through DDP to non-profit providers which deliver services in the communities.

To direct how income from the Trust is to be spent for each beneficiary, a Lifecare Plan is developed. This plan embodies the wishes of the donors and defines the scope and nature of supplemental services to be provided to the individual with disabilities. Trained Self-Sufficiency Trust counselors help donors to develop a realistic and need-specific plan. SST funds can be used to 1) maintain lifestyle by providing extras like leisure time activities, training, clinical services or transportation; 2) enhance opportunities for housing, supported employment or other government funded programs; or 3) provide life-long advocacy services.

In order to participate in Montana's Self-Sufficiency Trust, donors must do the following:

- Determine that the proposed beneficiary for an individual trust account is eligible for service provided by Public Health and Human Services (DPHHS).
- Register with MSST and pay the non-refundable registration fee ($100).
- Execute a transfer document. The transfer document should be prepared by an attorney or other qualified professional who represents the donor.
- Appoint an advisor for the individual trust account.
- Develop a Lifecare Plan.
- Have the Lifecare Plan approved by MSST
- Contribute cash or other assets to the MSST Private Fund.

The expense to the donor who is creating a Trust includes whatever costs are involved in executing their transfer document, the registration fee, and an annual management fee. The current manger of the Trust is P.A. Davidson in Great Falls.

Income from the MSST continues to be disbursed according to the dictates of the Lifecare Plan throughout the lifetime of the beneficiary. When the beneficiary of an MSST trust account dies, at least ten percent (10%) of the current market value of the individual's trust account goes into MSST's Charitable Trust, and the remainder is disbursed as designated by the donor.

The Montana Legislature amended the SST statute to make explicit that ten percent of the remaining principal must be designated to the Charitable Trust and the rest may be subject to garnishment.

Frequently Asked Questions About MSST

1. *What is a Medicaid-eligible trust?*

A Medicaid-eligible trust is one that has been approved by Medicaid as a trust to be used to supplement (not supplant) benefits provided by the state or federal government. The Montana Self-Sufficiency Trust is a Medicaid-eligible trust. Medicaid-eligible trusts may also be created on a private basis using the expertise of an attorney or estate planner.

2. *Can Medicaid require reimbursement from an SST account for Medicaid-funded services after the death of the SST beneficiary?*

The federal code governing Medicaid was recently changed so that Medicaid can claim reimbursement for Medicaid-funded services from the estate of a Medicaid beneficiary. Montana incorporated the federal code language into the Departmental Rules for Self-Sufficiency Trusts (Subchapter 5-45.2.502). The language in the Montana Rules says:

"The individual trust must provide that upon the death of the beneficiary the state be reimbursed, to the extent that monies remaining in the trust allow, an amount equal to the total cost to the state of providing Medicaid services to the beneficiary."

Practically speaking, this rule means that anyone who has received Medicaid services during their lifetime may have his or her estate garnished to reimburse the state for Medicaid services. An MSST account would have to be very large (i.e., several hundred thousand dollars) not to be fully utilized in reimbursing the state for services.

When the beneficiary of an SST account dies, whoever is the Advisor is obligated to notify the State of Montana that there is a remainder in the account, which can be used to reimburse the state for Medicaid services.

Montana law does have an additional provision, which says that ten percent of a remaining principal in an MSST must go to the Charitable Trust and may not be garnished to repay for Medicaid services.

3. *How should SST accounts be identified in order to comply with IRS rulings?*

An MSST account should be set up as a taxable entity with its own IRS tax ID number. In this way, the income from the trust for IRS purposes will be attributed to the trust account and not to the beneficiary individually. If the beneficiary is to remain eligible for Social Security, then he or she should not have any income, which would disallow government benefits.

The Trust Advisor will need to file an SS-4 Form (Report of Fiduciary Income) and a Form 1041 income tax return in order to comply with IRS regulations and to report the trust income in a legal way.

It is possible, though not particularly advisable, to set up an SST account under the Social Security number of the Donor or Trust Advisor. Then that individual would have to report the trust income on his or her regular tax return.

It is not wise to set up an SST account under the Social Security number of the beneficiary because the trust income will be attributed to that individual and may disallow the individual from receiving Supplementary Security Income and Medicaid.

4. *What is the current minimum for a deposit in an SST account?*

We recommend a minimum deposit of at least $2,500 because any smaller amount will be quickly "eaten up" by the annual management fee charged by Trust Corp.

5. *What management fees does Trust Corp charge?*

The Montana Self-Sufficiency Trust has an agreement with Trust Corp to manage the trust accounts. In the original agreement, Trust Corp said it would charge a minimum fee of $100. This minimum fee was really a charitable donation on the part of Trust Corp, since it did not cover their actual costs for managing the accounts.

As of June 1, 1996, Trust Corp has raised its management fee to more realistic numbers so that they can break even on their costs. On trust accounts with a personalized portfolio (individual stocks and bonds chosen by the donor), Trust Corp will charge a minimum of $500 as an annual management fee. For those accounts that deposit cash and agree to follow Trust Corp's recommended investment in the Federated Managed Series Trust, the minimum annual fee will be $250.

Any questions regarding fees should be addressed to Trust Corp at 1-800-634-5526. Copies of the Montana Rules governing the Self-Sufficiency Trust are available by calling PLUK at 1-800-222-7585.

To learn more about the Self-Sufficiency Trust, call PLUK and ask for the Donor and Attorney handbooks. These booklets will be mailed to you at no charge. ❑

Patient Realities in Genetics Testing

Contributed by Betsy Trombino

Editor's Note: Once viewed as a relatively obscure branch of medicine, genetics has become the focal point of diagnostic and therapeutic research that will eventually impact every facet of healthcare. With the scientific advances come greater burdens of responsibility for medical professionals and patients to make every effort to fully understand the ramifications of decisions affecting not only themselves, but virtually every member of a patient's family.

- Sally and Peter's baby boy, Jeffrey, is a year old and both the parents and their pediatrician have concerns about his development. Jeffrey is just beginning to sit up, crawl and eat solid foods. As part of the overall assessment, the pediatrician recommends a genetic test to check for chromosome abnormalities.
- Marian has just completed chemotherapy following a mastectomy for breast cancer. She has three daughters in their thirties and wonders about the possibility that the breast cancer is hereditary. She and her internist discuss genetic testing, but she wonders how to approach her daughters.
- Sherry is a young woman recently married. Her brother has Duchenne Muscular Dystrophy. She has asked her primary-care provider about the possibility of carrier testing before she and her husband consider having a baby.

These dilemmas are just three examples facing families and their physicians in the wake of the Human Genome Project and its discoveries. A routine diagnostic blood test can provide valuable information in treating a variety of diseases. A genetic test, however, goes far beyond the routine and has implications not always considered by the patient or the doctor.

Emotional impact. Often the results of a genetic test, particularly if they show that a person carries a "defective" gene, have a powerful impact on the basic premise of who we are as human beings. Such results may begin a process of grief that resembles other experiences of loss. How such information will affect a particular individual or family is unpredictable.

Family considerations. A genetic test usually does not involve just the person giving the blood sample. The results may have far-reaching implications for many family members, who may or may not be aware that the testing is even being done. It is imperative that these issues be discussed ahead of time and carefully considered by the patient and physician.

Financial implications. The cost of further testing and treatment of genetic disorders can be substantial, often far beyond what is covered by a family's medical insurance. Certainly this is not a reason to avoid testing, but it must be considered and funding alternatives identified as part of the decision-making process.

Insurance issues. The results of genetic testing have been used by insurance companies to raise premiums or even to cancel coverage. If an insurance company has paid for a particular test, they may be entitled to know the results and use that information to deny future claims. It is possible that the person/family who is tested may then be identified as having a pre-existing condition, making future insurability difficult, if not impossible. Again, upfront knowledge is imperative.

Future discrimination. Patients and their family members have reported discrimination in both work and insurance situations based on the results of genetic tests. Legislation at both the federal and state levels is addressing this issue, but patients and physicians need to be aware of the possibility before beginning the process.

Privacy and confidentiality. This issue exists on several levels, which need to be considered. Do other family members have the right to know test results, if they may be impacted? What about employers and insurance carriers? Who actually "owns" the blood/DNA sample? Can it be stored for future testing as new technology is developed? If so, who would have access to it? These are difficult ethical question with no simple answers.

Whenever a genetic test of any kind is recommended, physicians and their patients must consider all possible present and future implications of such testing, both for the patient and their family members. Since new discoveries are made almost daily, doctors and families will find themselves learning together. The more information each has, the better the chance that true informed consent will be achieved, and that a decision will be made weighing possible implications against the knowledge to be gained from testing. ❑

Social Security and Me

A Patient Perspective on Social Security

Contributed by Rod Slaght

Editor's Note: The patient experience in the management of a genetic disorder is rarely addressed, and poorly understood. Healthcare professionals often have no concept of the obstacles and frustrations patients face year after year as they work through the system to manage their personal, financial and medical affairs. The following article emphasizes the challenges patients must confront, and offers insight into areas of public policy and services delivery which must be addressed in the very near future.

My name is Rod Slaght and I am a person with a disability. I have been disabled since birth. The Social Security Disability System is so large at times it would seem to be unnavigable. I have had my share of ups and downs with the system. To an outsider it would seem they want to prevent you from succeeding. There appear to be many success stories, however data regarding long-term outcomes does not exist.

I was glancing through some papers that I picked up at one of the Mountain States Genetics Network's meetings called alphabet soup. This reminded me that Social Security has its own alphabet soup recipe. The letters spell out words like SSA, SSI, spenddowns, etc. If you visit your local Social Security office, they have pamphlets that explain each of these rules and the dates they were put into place. I would like to take a moment to touch on a few of these rules.

For individuals in the Social Security System, your assets may not exceed $2,000 and personal bank accounts cannot exceed a set dollar amount. Your car may not be worth more than $2,000. This last one I find humorous; keep in mind the rule was written in the 1970s. Today, what sort of vehicle could one find for that dollar amount that would be reliable? This rule prompted me to do some research. I found two sources that helped me in this task. One, a banking institution, and the other, a professional person who writes budgets for a corporation, both came up with the same dollar figure. If you were to find a comparable car that you purchased in the 1970s for $2,000, today it would cost you $4,132. This amount was arrived at by using a "very conservative" three-percent-per-year rate of inflation over the last 24 years. Social Security wants us to become self-sufficient and gainfully employed. How can one achieve this without a reliable vehicle? Many businesses will not even hire without personal transportation.

I grew up with a strong work ethic and being on the rolls of disability is beyond frustrating. For example, you may earn a limited dollar amount. Mine was in the $300-per-month range. This amount varies from person to person. If you went over the set dollar amount your SSI would be cut by the excess overage, be it ten, 20 dollars, etc. Another way to say this is you would have to repay $1 for every $2 you went over. At times when I worked for a fast-food restaurant, I was required to work over my hours. If I did not, a strong suggestion of being terminated was voiced.

Your choices are to leave the system completely or stay with the system as it is. I certainly cannot afford the medical insurance that exists outside the Medicaid system. It would be unthinkable to be without it. We all know how difficult it is walking into an interview for employment when you have a list of limited hours and monthly income. This reduces your chances of being hired.

Currently, I am in college achieving a management degree. After I graduate, I most likely will get help from the Rehabilitation Center and work on a PASS Plan to become employed again. A PASS Plan allows you to shelter your SSI or SSA benefits while you are re-entering the work force. Check with your local rehabilitation center for more details.

I am not alone. There are thousands of people like me with disabilities in this situation. I continue to write letters to my Congressmen and other elected public officials to try to educate them about our circumstances. As you can see, this is a difficult maze to navigate, and yes, beyond frustration.

There are some new rumors stirring from Washington that greatly excite me regarding these issues. The outcome is unknown at this time. We only hope for the best. ❑

Systems Advocacy

Providers As Advocates

Contributed by Doris Husted

Editor's Note: Ironically, as genetics permeates all areas of medicine, the impact of the medical, social, legal, ethical and economic ramifications will affect everyone. Changes in current systems must be made with the input and professional guidance of the medical community to guarantee that patients have the benefit of professional experience and knowledge working on their behalf as public and private healthcare policy evolves to accommodate the new medicine. Medical professionals truly are obligated to commit themselves to involvement in medical policy changes. Without that commitment, they will be as vulnerable as their patient to the effects of those changes on the practice of medicine.

"Who me? I'm a doctor! I have other things I need to be doing with my time. Besides, there are other people who are more qualified (better informed, more experienced, have more time) to do that."

Does this sound familiar? Every one of us, at one time or another has had this kind of a reaction. The reality is, however, that every moment of every day we are advocates. We advocate for ourselves; we advocate for our families; we advocate for our communities. Good citizenship requires

advocacy. Most people choose when they want to expand their advocacy efforts and which causes are worthy of their time and energy.

This ability to decide where to direct advocacy efforts is not available for individuals facing genetic testing or counseling, or who now have a genetic diagnosis. When we (or our child) receive a potential diagnosis or label, all of a sudden we are thrust into a whole new world. The world we thought we had is no longer under our control. The advocacy of physicians on behalf of both individual patients and the system is absolutely crucial. Every physician can and should be an advocate for his/her patient. The child patient won't reach his full potential; the individual may not even live if doctors don't don the role of advocate.

Policy about the direction and role of genetics is also being made by public opinion and private entities without the input of the public scientific community such as those research groups funded with federal dollars and individual practitioners. Every man, woman and child will feel the impact of the policy which results. These discussions and decisions need to remain in the public domain.

Advocacy takes place at three levels: systems advocacy, individual advocacy, and personal or self advocacy.

Systems advocacy is intended to change the system in which healthcare is provided. State insurance offices, state health offices, state legislatures, Congress and the U.S. Department of Health and Human Services are only a few of the arenas for systems advocacy. Each of these entities has some level of control over what healthcare is provided, how that care is delivered and, maybe most important, what will be paid for and by whom. It is very important that physicians be involved at this level. They have the ability to explain the consequences of decisions (pro or con) in patient-specific terms and the implications for society as a whole.

Cloning is but one example of the need for systems advocacy. The broader impact of prohibiting cloning has received very little or no coverage in the media. All the reports have been about the cloned animal and how humans will be next. Doctors could and should be talking about the treatment of people who have been severely burned and what will happen to their treatment if cloning is banned.

Individual advocacy is time and energy spent advocating on behalf of an individual. Every physician does this over and over again in the practice of medicine. Each patient's needs are different and many times it takes the doctor's phone call or the doctor's letter to assure the patient receives the necessary attention, whether it be tests or treatment. Patients must have faith that their doctor is willing to make waves on their behalf, not just receive payment for services rendered.

Personal or self-advocacy is the time and energy spent on personal concerns. Physicians are also people with families and issues that need attention.

No matter where we choose to spend the majority of our time in advocacy - systems, individual or personal - it is the fact that one is doing advocacy which is of prime importance. Physicians and other professionals must be at the table when decisions affecting genetics and other healthcare services are made for their patients, for their practice, for the future. ❑

Genetics and Managed Care

Contributed By Susan M. Bryan

As a species, we have always been obsessed with our own health and forestalling our own demise. Medicine is the manifestation of that obsession and for many, medical genetics is the perceived solution to conquering sickness, and even death.

But while genetics may be the wished-for panacea of science and medicine—not to mention big business—our quest for a healthcare utopia is rapidly leading us to a social purgatory, because the moral, legal, ethical, political and economic ramifications of the genetics revolution are only beginning to manifest.

The complex issues surrounding genetics go beyond the obvious impact on healthcare methodology, demanding examination of many factors. But beyond the obvious direct impact on healthcare, genetics will also change the economics of medicine. As developments in the use of genetic technology increasingly impact the practice of medicine, healthcare providers and public and private systems will be challenged to find resources to meet the needs of the population.

As more information from the Human Genome Project is refined and applied to new standards of care, virtually every area of medicine will be affected. New diagnostics and new therapies could potentially be applied to every individual in the population – even to those not yet conceived. Yet to actually provide these services, delivery systems and reimbursement policies will need to change radically. Direct and indirect costs will stretch private and public resources, and the new medicine may well become the key factor instigating a compete revamping of the current managed care system.

The true challenge of the new medical genetics may well be determining how to pay for it. How much money will be needed to provide genetic tests, therapies, education and counseling for every single member of the population?

Patient demand and commercial gain will be the driving forces behind the new medical genetics. In a perfect world, all medical services would be available on demand to all members of the population. But existing and near-future reimbursement protocols do not support widespread provision of genetic services. Last year in the United States, more than *one billion dollars* was paid by third party and individual consumers to cover existing medical services, including the limited genetic services available. In perhaps 10 years, what will the cost be of new medical services based on the potential genetic testing and treatment of the entire population?

Of course, widespread application of genetics in everyday medicine is still in the future, and current healthcare coverage is based on patient outcomes and relative benefit. Today's outcomes model supports the extension of life, some preemptive (wellness) care, and general coverage for accepted standards of care, most of which include only the minimum genetics services associated with prenatal and some cancer diagnostics.

To date, relatively few outcomes analyses of the cost of clinical genetics have been developed or published. For most of the more recent genetic tests and treatments, there has not yet been enough time or usage to apply the

standard algorithm of research, practice and policy development which is normally used to change standards of care.

Despite this reality, it is unquestioned that genetics will alter all standards of care rapidly. The costs for much of current genetic testing are covered by Federal and State-supported programs. But as additional genetic services become available, and as the population increases and ages, public health budgets will be further stressed by rapidly expanding Medicare and Medicaid populations in need of both short and long term care.

There are no easy answers to any of the logistical or fiscal (not to mention the social, ethical and political) issues surrounding provision of genetic services. But the inescapable reality of advancing medical genetics options requires launching full-scale efforts to resolve as many questions as possible before the potential for genetic medicine is fully realized. Several states have been reviewing the question, with initial plans for educating the public and delivering some genetics services in place in Arizona, Hawaii, Ohio, Texas and Washington. Colorado is in the process of assessing current and projected need and developing programs to educate its population regarding the present and future options in genetics. The final plan will need to provide public health strategies for the education and support of genetics services providers, primary care providers, managed care and consumers in meeting the challenges of providing genetics services in a rapidly changing medical environment. These strategies must be appropriate for the long-term, taking into account a system that will continue to evolve, and the costs and needs of the required support systems that will sustain optimum services for the public.

At this juncture, we can be futurists, idealizing the potentially glowing opportunities presented by the advances in genetics. But as appealing as that may be, logic demands that we first examine the future potential and align it with the needs and realities of the present. The future will then take care of itself. ❑

Top Ten Reasons Medical Professionals Should Be Consumer and Patient Advocates

Contributed by Susan Duffy

As consumer activists working to support patients challenged with genetic disorders, we strongly urge those of you who are involved with planning professional conferences to actively solicit and support consumer participation at those events. To be frank, this means providing stipends to make it financially possible for consumers and patients to attend. What we have to offer, however, makes us cheap at twice the price. In the spirit of David Letterman, therefore, we offer medical professionals the top ten reasons for providing funds to bring consumers to your conferences.

10. It will make you look golden on your grant applications.
9. We need the relief from the reality of dealing with the challenges of patient issues.
8. Many of us can offer you related expertise from our own professions such as journalism, education, law, economics and other diverse fields.
7. At some time in your life, you may become a patient or "consumer" yourself, and we are truly useful people to have in your Rolodex when you need to network from that "other" perspective.
6. Often we are politically well connected, and we make terrific advocates at legislative and congressional hearings.
5. We have the best (most shocking, most heart-rending, not to mention funniest) anecdotes you have ever heard.
4. As a defense to reality, we have refined black humor to an art.
3. You, along with us, can use the opportunity to show each other that we are far more than that bloodless "professional" label and its oh-so-passive "consumer" equivalent.
2. Face-to-face meetings are the most likely to lead to positive movement on most issues. This is why nations hold summit conferences, right?
1. We really are all in this together, so let's *get* together and combine strengths. (And remember those grant applications...) ❑

The Mountain States and the Delivery of Genetics Services

Contributed By Susan M. Bryan

As stewards of public health, state health programs are increasingly challenged by the growing need for information and services related to genetics. Due to increasing media attention to issues relating to the Human Genome Project, identifications of genetic markers related to various disorders, and the creation of new diagnostic and therapeutic procedures utilizing information gleaned from genetic research, the public has a new and intense interest in learning how genetics can positively impact their health.

Unfortunately, conflicting messages in the media and limited information from traditional health resources can result in consumer confusion and frustration. In order to fully understand the impact that genetics can have on personal health, patients and their families may best be served by looking to their state departments of public health. As part of the general structure of the states' programs, genetics coordinators head up various programs, which receive federal, and state funding to provide the population with referral services, educational materials, and sometimes networking opportunities among medical professionals and consumers.

The states are also looking toward the future, as the issues regarding personal genetic information, privacy, ethics, and reimbursement become more complex. Many states, including Arizona, Colorado, Hawaii, Texas, and Washington, are assessing future populations needs and equal distribution of services and education, developing in-depth programs which will serve all individuals and guide healthcare services providers as they work to meet public demand. Changes in medicine are expected to necessitate changes in public policy and private practice which will significantly alter healthcare in the future. These plans are being developed in advance of what is expected to be a revolution in the detection, evaluation and treatment of disorders, illnesses, and physical problems through genetics.

Currently, Arizona, Colorado and Utah have a state genetics advisory committee. The state programs generally follow the model under development in Colorado. There, a State Genetics Advisory Committee made up of representatives from private and public genetic centers, the state health department, universities, consumers, private laboratories, HMOs, private legal practice, and religious and ethical review boards are assembled to discuss assessing need, developing policy, and assurance of parity in the provision of information and services.

Models to meet all possible needs of the public in these areas are as much in their infancy as genetic medicine itself. Yet the rapid advances in the field and the extraordinary promise of genetics in the treatment of congenital and acquired disorders ranging from inborn errors of metabolism to cancer are driving research and application at astonishing rates. State health departments and special review boards like Colorado's State Genetics Advisory Committee will be required to promote family-centered, community-based, culturally competent, comprehensive, coordinated social and health care systems of clients and families to meet genetic health care needs.

To meet this directive, the input and insight of representatives from all sectors of the population will be critical. With this in mind, medical professionals and consumers alike are encouraged to contact the coordinators in their states (listed below) to become involved in the review and program development process. ❑

State Genetics Coordinators

Arizona (Acting)
Karen Kuhfuss
Arizona Department of Health Services
411 North 24th Street
Phoenix, AZ 85008
Phone: (602) 220-6343
FAX: (602) 220-6551

Colorado
Joyce Hooker
Medical Consultation and Genetic Services
4300 Cherry Creek Drive South
Denver, CO 80246-1530
Email: joyce.hooker@state.co.us

Montana
John Johnson, MD
Shadair Hospital
P.O. Box 5539
Helena, MT 59604
Phone: (800) 447-6614
FAX: (406) 444-7536
Email: jjohnson@initco.net

New Mexico
Sue Brown
New Mexico Department of Health
P.O. Box 26110
Santa Fe, NM 87502
Phone: (505) 827-2354
FAX: (505) 827-1697

Utah
Lynn Martinez
Utah Department of Health
Salt Lake City, UT 84114-4691
Phone: (801) 534-8541
FAX: (801) 584-89[not legible]

Wyoming
Larry Goodmay
Genetic Program Manager
Wyoming Department of Health
Hathaway Building, 4th floor
Cheyenne, WY 82002
Phone (307) 777-7166
FAX: (307) 777-5402

Resources

The following resources are available for more information regarding genetics and healthcare in the Rocky Mountain States. This is not a complete listing; for more information, visit the Mountain States Genetics Network website at http://www.mostgene.org/

The Family Health Library at
The Children's Hospital
1056 E. 19th Avenue
Denver, Colorado 80218
Librarian: Kate Smith
Phone: (303) 861-6378
E-mail: family.resource@tchden.org

University of New Mexico Health Sciences Center
Children's Hospital of New Mexico
Family Resource Library
Librarian: Julia D. Grimes, M.L.S.
Phone: (505) 272-1707

Genetic Alliance
4301 Connecticut Avenue NW, #404
Washington, DC 20008-2304
Phone: (202) 966-5557
(800) 336-GENE
Fax: (202) 966-8553
E-mail: info@geneticalliance.org
Website: http://www.geneticalliance.org

Mountain States Genetics Network
Colorado Department of Public Health
4300 Cherry Creek Drive South
Denver, CO 80246-1530
Phone: (303) 692-2423
Fax: (303) 782-5576
Website: http://www.mostgene.org

National Human Genome Research Institute
National Institutes of Health
9000 Rockville Pike, 49/3A14
Bethesda, MD 20892-6050
Phone: (301) 496-0844
Fax: (301) 402-0837
Website: http://www.nhgri.nih.gov

National Organization for Rare Disorders (NORD)
P.O. Box 8923
New Fairfield, CT 06812-8923
Phone: (203) 746-6518
(800) 999-NORD
Fax: (203) 746-6481
E-mail: orphan@rarediseases.org
Website: http://www.rarediseases.org

Office of Rare Diseases
National Institutes of Health
31 Center Drive, MSC 2082, Room 1B03
Bethesda, MD 20892-2082
Phone: (301) 402-4336
Fax: (301) 402-0420
E-mail: sg18b@mh.gov
Website: http://www.rarediseases.info.nih.gog/ord/

Colorado Department of Public Health and Environment
Medical Consultation and Genetic Services
4300 Cherry Creek Dr. South
Denver, CO 80246

Director—(303) 692-2424

Coordinator—(303) 692-2423

http://www.mostgene.org

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