ASHG 1999 PRESIDENTIAL ADDRESS:
Human Genetics in the Information Age
Uta Francke

Ladies and gentlemen, members and guests, colleagues and friends!

It has been a great honor for me to serve as the president of our Society this year.

The purpose of the society is to bring together investigators in many areas that involve human genetics, and to encourage and integrate their efforts by providing a forum for sharing research findings. This is accomplished primarily through the Society's annual meeting and our monthly publication, the American Journal of Human Genetics. With a focus on education, ASHG strives to inform health professionals, legislators, health policy makers and the general public about all aspects of human genetics.

During this year changes have occurred in all three areas: the meeting, the journal and public involvement.

One of the few things I have been able to influence, is to reorganize the program of the annual meeting. The invited speaker sessions, previously divided into symposia and workshops, were always clustered on the first day. In the future, they will be spread out during the entire meeting. The invited speaker sessions, presenting coordinated reviews of timely topics, constitute the major educational component of the meeting. Each member of the society is asked to contribute ideas for topics, organizers and speakers to the program committee.

As part of the re-organization, the official opening of the meeting with the presidential address and the first plenary session was moved up to the first day. The program committee also agreed to add a second plenary session, so that each of us can hear more of the exciting new work in areas outside of our own subspecialty.

Our society has over 6,700 members, a 3% increase from last year. It is truly international, with more than a thousand members living overseas. Demographic information, collected over the last two years, reveals great diversity. About half of the members are engaged primarily in research, 30% in clinical work, 10% in counseling, followed by administration and teaching. Obviously, most of us combine several of these activities. Of members with MD degrees, half are engaged primarily in clinical work, and 65% of Ph.D.s are primarily in research with 21% in clinical labs.

The settings of laboratory-based work are 50% in university research labs and 42% in various diagnostic labs. For those that do research, the type ranges from basic or applied clinical to basic mathematical research.
This level of diversity sets our society apart from other learned societies that are either oriented more toward the basic research or more toward the clinical/applied side. We are a true hybrid. Our meetings and our journal provide opportunities for learning and interaction across a wide range of human genetics research.

The diversity also presents challenges. One is to discourage special interest groups from walking out and forming their own societies. In Europe, for example, the formation of a regional cytogenetics society has left the European Society of Human Genetics somewhat anemic, as judged by attendance at the last annual meeting. I hope that you all value the opportunities of belonging to a large diverse society; that you'll feel at home at our meetings; publish your best work in our journal and consider taking part in the many activities that ASHG committees are devoted to throughout the year.

Earlier this year, the society was admitted to full membership in FASEB - the Federation of American Societies of Experimental Biology - which is the nation's largest association of biomedical scientists. We have had a long relationship with FASEB. They provide logistical support and space for our administrative offices in Bethesda, as well as organizational support for this meeting. The decision to apply for FASEB membership had been postponed over the years and I am proud that it was achieved this year. Made up of 19 Societies, the Federation represents an aggregate membership of 63,000 investigators. It has a collective voice on matters of legislative and public interest and campaigns actively for biomedical research funding. ASHG now has representatives on the FASEB Board of Directors, and on various committees, including public affairs and science policy.

I will not limit my talk to telling you statistical and organizational details. I rather wish to share my thoughts on a timely and controversial topic:

• the information revolution and its impact on human genetics.

The themes of change and challenge, even evolution, have intrigued previous presidents, as you can see from the titles of their talks:

Table 1: ASHG Presidential Addresses

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<tr>
<th>Year</th>
<th>President</th>
<th>Title</th>
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<tr>
<td>1992</td>
<td>Walter Nance</td>
<td>Back to the future</td>
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<tr>
<td>1993</td>
<td>Janet Rowley</td>
<td>Can we meet the challenge?</td>
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<td>1994</td>
<td>Maimon Cohen</td>
<td>Who are we? Where are we going?</td>
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<td>Anticipating the 21st century</td>
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<tr>
<td>1995</td>
<td>Judith Hall</td>
<td>The challenges and opportunities of times of change</td>
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<td>1996</td>
<td>Charles Epstein</td>
<td>Towards the 21st century</td>
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<td>1997</td>
<td>Larry Shapiro</td>
<td>Genetics, genomics and geneconomics:</td>
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<td></td>
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<td>Market forces and natural selection</td>
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<td>1998</td>
<td>Art Beaudet</td>
<td>Making genomic medicine a reality</td>
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At this 49th meeting in the year 1999, we stand on a threshold - with big round numbers
coming up next year. It is, therefore, appropriate to look back from where we came.

One of the messages I wish to convey is that

- **professional life is a constant learning process, you never cease being a student**

I will illustrate this point with my own experience. As a foreign medical school graduate, I started out in pediatrics. When I entered human genetics, 30 years ago, chromosomes were merely counted, they could not be identified. During my first year of genetics fellowship, chromosome banding methods were developed. They allowed us to define new chromosomal syndromes and map genes to individual chromosomes and regions.

With the advent of somatic cell hybridization methods, human disease phenotypes could be studied in cultured cells. Selection methods and complementation tests were devised - following strategies of bacterial genetics. When we were able to make interspecies somatic cell hybrids, gene mapping took off in the 1970s. In the early 1980s, recombinant DNA techniques enabled us to map genes by FISH and restriction fragment analysis.

In the mid-1980s, PCR was invented and completely changed the ways of experimental research. In my lab, standard microscopes were packed up and put into closets.

They were replaced by PCR machines and a confocal laser microscope with a computerized imaging unit. Automated DNA sequencers were added shortly afterwards, as we progressed from mapping to sequencing. The Human Genome Project then moved the work out of academic research labs to large Genome Centers, and we focused our research on the molecular genetics of specific disorders.

Ten Years ago, I moved my lab from Yale University to Stanford Medical School into the Center for Molecular and Genetic Medicine. The brand-new laboratories were state of the art and, curiously, each desk in the lab had a round hole in it. The idea was that, eventually, computer cables would be routed through these holes. At that time, we had one computer for the entire lab. Well, it didn't take long for computers to appear, one on each desk. Whenever you go into the lab now, you will see at least half the people working in front of the screen rather than at the bench.

- **The revolution in information technology is changing the way we do our work - and even the type of work we will be doing - in research, laboratory diagnostics as well as clinical genetics and counseling.**

Before I stick my neck out predicting the future, I will share with you a few historical vignettes that I clearly remember.

In 1977, when John Opitz started the American Journal of Medical Genetics and I was enthusiastic about participating as an Advisory Board member, my Human Genetics department chairman thought this venture was unnecessary; the esoteric specialty of Medical Genetics did not need another journal. In the two decades since, all of you have witnessed the
growth and professionalization of our specialty.

In the mid-1980s when I was on the Board of Directors of the American Board of Medical Genetics I urged the establishment of a specialty board in Molecular Genetics. The chair of the Board rejected this idea by saying: Molecular genetics is just a method, like electrophoresis, that will be used by everybody; it is not a career path. In 1990, molecular genetics was added to the biochemical genetics board exams and, ultimately, in 1993, Molecular Genetics Boards were instituted.

In last year's polls (reflecting responses of 94% of the ASHG membership) molecular genetics was chosen as the major category under "Special areas of interest" by nearly 2000 people, followed by cancer genetics, prenatal genetics and cytogenetics. The 1999 data are almost identical.

**Bioinformatics** was not one of the options on this questionnaire. In recognition of the increasing importance of this field, informatics has been added as a "Special interest area" to the current Member Profile form.

I will now consider the impact of the information revolution on 3 areas that are tightly linked to each other:

**NEW KNOWLEDGE:**

**Acquisition - Dissemination - Interpretation**

How we generate new knowledge through our research
How we disseminate it through various means of publication
How we interpret it for the general public and for the unique situations of our patients

**I. Acquisition of new knowledge**

We have witnessed the move to global approaches, from mapping one gene at a time to sequencing an entire human chromosome to sequencing the entire genome. New technology is speeding up progress and a first draft of 90% of the human genome sequence is expected to be publicly available next Spring.

What are we going to do with that string of a four letter alphabet; 3 billion units - half as many as there are people inhabiting the planet? (We will know where everybody lives, though, because the sequence will be linked to the chromosomal position)

This is where **bioinformatics** comes in. Its function is to extract knowledge from data. More precisely, bioinformatics is the process by which data are gathered, organized and computationally analyzed. Bioinformatics can be viewed as a theoretical approach to biology that makes use of computer algorithms and existing knowledge to mine nuggets of useful information from the mass of molecular data available.

Genes will be identified by computer programs rather than experimentally. Their function
will be deduced by comparison with functional information derived from experimental organisms that have undergone complete genome sequencing and systematic functional analysis of all their genes.

Systematic functional analyses of all human genes will be approached through global expression analyses of normal and diseased tissue by microarray hybridization. Once the genome is sequenced and all human genes are identified, high density microarrays containing all genes will allow the collection of large data sets with relatively little effort.

Pat Brown, a pioneer in this field has said: "Experimental science has been liberated from the dogma of hypothesis-driven research to the new freedom of exploratory research". The lets-do-it-and-see-what-we-find approach - usually disapproved by grant review committees as a "fishing expedition" - is becoming fashionable. Why is that? The technological fishing nets are such that the catch is expected to be valuable - no matter what it consists of. The catch will be large, for sure, but the gold fish may be hidden in a school of herring.

Anyone who receives the results of a gene expression microarray analysis, even containing only a portion of all human genes, is impressed by the daunting task of making sense of the wealth of information. Here, the tools of bioinformatics are essential. Cluster analyses reveal groups of genes with similar expression patterns. These clusters are linked to biological processes; they can be developmental stage and cell cycle specific, cell type and tissue specific, disease process-specific - endless categories will be recognized and defined.

This kind of global exploratory research with massive parallel measurements will generate new hypotheses and predictions that then require specific experimentation to be tested:

- Unknown genes will be discovered with functions predicted by their sequence.
- Known genes will be predicted to have novel functions based on expression data or cross-species homology data.
- Genes will be predicted to be involved in specific diseases

To give you an example: As we published in the October issue of Nature Genetics and Huda Zoghbi will present in the late-breaking research session on Friday morning [1, 2], Rett syndrome - a sporadic brain disorder affecting young girls - is caused by mutations in the MECP2 gene. The gene is on the X chromosome and encodes a protein that binds to methylated DNA and is part of a silencing complex that modifies chromatin structure and shuts off transcription.

To understand how mutations in this gene cause the neurodevelopmental phenotype we need to know the specific target genes that are dysregulated. Gene expression microarray analysis is used - in collaboration with Pat Brown's lab - to identify transcripts that show quantitative differences in mutant cells versus controls. This approach will not provide definitive answers. It is only the first step in identifying possible candidates as targets for MECP2 regulation that will need to be examined by focused experimentation.

The public availability of the human DNA sequence will enable the concentrated (and
competitive) effort of mining it for genes that are responsible for or predispose to diseases and encode potential drug targets. What will we need to be successful?

We will need an **organized interdisciplinary effort** that includes

- **mathematicians and software engineers** to further develop the existing tools for comprehensive sequence analyses, for identifying coding genes and functional domains, for defining gene families and evolutionary relationships; for predicting three-dimensional structures of proteins and protein-protein interactions based on known structures and interactions that were experimentally determined; and for developing integrated front ends that make it possible for non-computer experts to move around online databases with ease;
- **biologists** to formulate the questions to ask the computer experts, to interpret the output and to annotate the sequence;
- **clinicians and pathologists** who understand disease processes and outcomes to suggest possible links between genes and diseases.

Data by themselves are useless, unless they are turned into knowledge by the interaction of all these components.

The components of such a multidisciplinary effort are represented by people who have the appropriate training and experience, ideally, people trained in more than one field who can speak and understand the language of another field. You can get there by dual training if you are young, or by cross-training if you are in one field and want to bridge into another. In the future, training in bioinformatics will be required for any biology or medical student, just as we now require that they learn organic chemistry.

Bioinformatics teaching, tools and search engines are developed by government agencies such as the **National Center for Biotechnology Information (NCBI)** [http://www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov). Its mission is to organize DNA sequence and related data in such a way that it is available for investigators to use and analyze.

**PubMed** is a system designed by NCBI for searching MEDLINE, the database of biomedical abstracts [http://www.ncbi.nlm.nih.gov/entrez/query.fcgi](http://www.ncbi.nlm.nih.gov/entrez/query.fcgi). It has recently changed its scope by including new features; one is called Coffee Break, a collection of short reports on recent biological discoveries. Each report incorporates interactive tutorials that show how bioinformatics tools are used as a part of the research process.

There are also products of commercial companies that provide integrated access to numerous diverse data bases and can tailor the search and analytical tools to individual user's needs [e.g. http://www.genomica.com; http://www.doubletwist.com](http://www.doubletwist.com).

- **New knowledge will be created by generating new links between existing information.**

In the case of Rett syndrome, a wealth of data existed on a specific gene and its function, the methyl-CpG binding protein MECP2; another pile of information existed on the specific
disease, e.g. information on sex-limited occurrence, stage-specific manifestations, pathological findings, population frequencies, exclusion mapping data. To identify MECP2 as the Rett gene, someone had to establish that link.

In the future, sophisticated programs will need to be developed that integrate all existing information on genes and heritable disorders to narrow the number of candidate genes and the number of diseases for a possible match. Ultimately, knowledgeable clinicians with a hunch and/or massive re-sequencing efforts will be successful in establishing a link.

I believe there is an opportunity for clinical geneticists, developmental biologists and others to participate in annotating the human genome sequence, in mining it for functional relevance and matching of genes with disease profiles.

II. Dissemination of new knowledge

Publication in peer-reviewed printed journals is the current standard. Journals fall into two categories: commercial for-profit owned by publishing houses and not-for-profit owned by learned societies, like the American Journal of Human Genetics (AJHG). The move from exclusively paper journals to electronic versions of the paper journals is progressing rapidly, with new electronic-only journals also being launched. Society-owned journals like ours are freely distributed to the members. As long as you pay your membership dues, you'll get the journal in the mail each month and you have free online access to a site where you can read accepted papers that will appear in a future issue in print [http://www.journals.uchicago.edu/AJHG/index.html].

How many people are actually reading or downloading AJHG articles?

[Thanks to Bob Shirrell, U. Chicago Press, for the web user data]

A user session is defined as one log-on session, regardless how many issues or papers are
viewed or downloaded. There was a rise in early Spring when the paper journal was late in coming out, and now the numbers are steady around 5000 per month.

Will paper journals disappear? Maybe, after a transition period \([3-5]\). And here is why:

[from Walker TJ, 1998, ref. 5]

Look at the way we have been publishing our work. We electronically generate a manuscript, submit and revise a printed version. Then thousands of copies are printed on a central machine, a journal is bound, wrapped, address labeled and mailed to libraries. Here the package is opened, the journal inventoried, labeled and shelved. It is then picked up by a researcher and taken to the copy machine to generate a paper copy of variable quality depending on the state of the copier. Alternatively you can request reprints, that are paid for by the author, who labors on mailing them out. This is the way we lived for decades.

Here is the way of the future: An electronically generated manuscript with associated figures, tables and data is electronically submitted to editors, is electronically reviewed, revised and put on a web server for everybody to read. Less time has passed and not a single tree had to die. If you find it easier, as I do, to read printed copy than a lighted screen, or want to take a copy to the beach, you may sacrifice part of a tree and print out a clean version on your own printer.
Besides being economical and fast, electronic publishing has innumerable advantages. Printed journals have limited space; pages cost money. Electronically published manuscripts have no such limitations; cyberspace is unlimited and it offers creative novel formats for publications [4].

For example, I can envision multi-layered structures with the first level an extended abstract - for the superficially interested reader; the second level has expanded figures and documentation; and finally, there are original results of experiments and extensive data files such as genotyping data of a linkage study or expression microarray data - for people who are interested in the details or want to conduct a meta-analysis. Most excitingly, video clips of movements in time-lapse or real time can be included. All life processes are dynamic. Movements and behaviors of cells, animals and human study subjects can be captured on video and made part of electronic publications. Furthermore, a comment box could accompany each paper in which readers can place confirmatory or critical responses. Rather like reviewers' comments about books on amazon.com.

So, let's say all existing journals will go online and new e-journals taking advantage of the interactivity and connectivity of the internet will be started - is that all we need for scientific communication in the next century? No, emphatically no. What we also need, is free and unlimited access.

When you read a paper in the online version of the AJHG and you want to check out a reference, you can click on it and get linked to the MEDLINE database of published abstracts. That's free and it may be all you wish to see. But if you want to read the full paper you may run into a wall [3,5]. To access the full text you may be asked for a password. Only subscribers get passwords. You can buy an annual subscription and get immediate online access to one journal only. Or you can charge your credit card for the right to read one article.

When you walk over to the library to search for the article and copy it, you may discover that your library has discontinued its subscription to that journal. The cost of library subscriptions have skyrocketed in recent years to the point that universities cannot afford to keep all subscriptions they used to carry or take on many new ones.

University libraries are in a financial crisis. The increase in journal costs to libraries is due to for-profit commercial publishers who took over the majority of scientific publishing when the learned societies and university presses could not keep up with the flood of manuscripts produced. The publishing company Elsevier that has just bought the Cell Press journals owns 8 of the 10 most expensive biomedical journals. Some of these carry library subscription price tags as high as 15,000$ per year. For comparison, a library subscription of our society's journal costs 525$. Commercial journals and some learned society journals, like the New England Journal of Medicine and Science, make sizable profits based on their extensive advertisements alone, yet they restrict access to the papers they publish.

Consider that the research results which are communicated in the journals are generated with support of taxpayer's money, either directly through NIH/NSF or DOE grants or indirectly through foundation grants. Therefore, the unimpeded dissemination of these research results
is in the public interest.

Consider further that the manuscripts and the peer reviews are contributed free of charge by us the scientists - why should we continue to put up with commercial publishers that erect firewalls to restrict access and collect profits?

On top, you see how the current system works: A publisher takes care of the reviewing, editing and composing of your article and puts it onto a commercial web server. Access is limited to subscribers (S), staff at institutions that purchase site licenses (SL) and people who charge their credit card for reading one paper (PPV pay per view). These three mechanisms generate income for the publisher.

Under a free access model, the publisher would still do the reviewing, editing and composing, but this would be paid for by the authors and their institutions who would save money by not having to pay for subscriptions and site licenses. It has been proposed that some of the university library budgets could then be channeled to departments to help faculty members pay for the publication of their work. Grant application budgets already have publication costs as a line item to pay for color reproductions, page charges and reprints that can be re-dedicated to pay for electronic publications. Just as print journals are full of commercial ads,
electronic journals can be supported in part by advertising income.

The existing system has received a major challenge earlier this year when Harold Varmus, Pat Brown and David Lipmann proposed the establishment of e-biomed, an electronic repository for biomedical research papers that would be distributed freely through a web server at the NCBI. [http://www.nih.gov/welcome/director/ebiomed/ebi.htm#Addendum] It was modeled after a very successful electronic preprint server for the physical sciences that was established in 1991 at the Los Alamos National Laboratory.

After heavy debate and feedback from the community the e-biomed proposal was modified. A few weeks ago, Varmus announced that the new venture, called PubMed Central, will be an archive of primary research papers in all areas of life sciences, not limited to biomedicine. It will be integrated with PubMed the search engine for the MEDLINE database of biomedical abstracts. It is envisioned that all journals will eventually participate and deposit their issues after print publication.

The Board of Directors of our society has agreed to consider participating in PubMed Central after it becomes operational early next year. Whenever that decision will be made, the work that you publish in the journal will be freely available online and will be electronically linked to the biomedical literature 2-3 months after its print publication. The advantages are numerous. Scientists in other fields, who wouldn't pick up an issue of AJHG and browse through it, will be made aware of our work through internet links to specific papers. This will expand the readership and foster the connection and collaboration of human geneticists with scientists in other areas. This development will accelerate the discovery of new knowledge and its worldwide dissemination.

The success of PubMed Central will depend on how many journals are placed into the repository and the extent to which old volumes that were only produced on paper can be converted and deposited. Other learned societies with high impact journals like the American Society of Cell Biology have agreed to participate. PubMed Central will also consider research contributions that are not published in any print journal as long as requirements for some kind of screening or review are met. You can read the details of the plan at [http://www.nih.gov/welcome/director/pubmedcentral/pubmedcentral.htm]

Ultimately, the growth and continued life of PubMed Central will depend on us - the authors of scientific papers. We have a choice where to submit our work for peer review and publication: to a journal that will make it available free for all on the web or to a journal that will use the copyright to embargo its free distribution. As authors and consumers, we have power and influence over the developments in electronic publishing.

As members of the human genetics community, we have opportunities and obligations to facilitate the publication of high-quality genetics research - by serving as reviewers and editors for existing journals, as well as for electronic journals yet to be founded. If we want free access to the scientific literature online, we can make it possible. We do have a choice to which type of journal, PubMed Central contributor or restricted access type, we dedicate our unpaid labor of peer review. Ultimately, the quality and speed of peer-review and editing will
determine which journal gets the best manuscripts. Quality of manuscripts, in turn, determines the journals' citation ranking, impact and prestige. The current hierarchy is not set in stone.

III. Interpretation of new knowledge

If we were entirely focused on the acquisition and dissemination of new knowledge and on communication with our colleagues, we would completely miss the boat. One of the most striking features of the Information Age is the involvement of the general public, including the education system, our patients and their voluntary support group organizations. By accessing web-based information, our patients are better informed than ever and their organizations are able to spread new knowledge with lightning speed.

Here is an example:

When the gene for Rett syndrome was discovered recently [1], Kathy Hunter the founder and president of the International Rett Syndrome Association (IRSA) designed a list of questions that families of affected girls might ask: What does this mean? Will all affected children have the same mutation? Where can my child be tested? How close are we to treatment, to a cure? How will a mouse model help? She had her written answers checked by scientists and then posted the document on the organization's web site [http://www.rettsyndrome.org].

On the site, she also copied the news releases issued by the institutions that had supported the research. Access to the paper itself was, of course, restricted to subscribers of Nature Genetics, but the journal allowed the commissioned News and Views article about the discovery to be placed on the IRSA web site. As soon as the news embargo was lifted, Ms. Hunter sent an e-mail message to the thousands of IRSA members telling them "The Rett gene has been found - visit our web site to learn more". This strategy not only protected Ms. Hunter and others from becoming overwhelmed with phone calls and e-mail messages full of questions, it leveled the playing field by giving everybody around the world access to the same information at the same time.

This example illustrates the attributes of web information: it is democratic, global, instantaneous, and free (except for internet access and the use of phone lines). The division line between HAVEs and HAVE NOTs is drawn by internet access. Only 5 years after Netscape released their first browser, 40% of Americans are internet users and this percentage is higher in the younger generation. It will not take long for the rest of the world to catch up, possibly using signal transmission via satellites.

Health information on the web is in a rapid growth phase. Commercial sites provide searchable information on any health-related topic, with hyperlinks to disease-specific sites, literature and other resources. They are supported by advertisements and, therefore, offer free access, while encouraging you to buy your drugs and medical devices online. Before you can search for information, you are asked to register and provide your e-mail address. If you list any diseases or topics you are interested in, you will be notified by e-mail of relevant new
developments.

In addition to information for consumers, WebMD.com offers full services for health care providers from office management to online stock trading; there are chat rooms on specific medical topics, lectures by experts in the virtual WebMD auditorium, and tests you can take for CME credit.

Everett Koop, a former US surgeon general, maintains a site with health-related news and a large searchable encyclopedia [http://www.dr.koop.com]. He also evaluates and rates other health related web sites.

At Americasdoctor.com you can find out about clinical trials that you may want to participate in. You can also speak to a doctor 24 hrs a day. There are real physicians sitting at terminals linked to databases who will answer your specific questions in real time with no charge. To summarize the disclaimer you have to agree to first: These physicians will help you in your search for information, but they will not engage in a doctor-patient relationship with you. Whatever they do for you, does not constitute "practice of medicine". This disclaimer is a legal necessity because the licensing of physicians is controlled at the state level. Well, in reality many doctors may be less informed about a specific subject than these internet docs sitting at their terminals with the latest information at their fingertips. And sadly enough, most doctors in practice today have very limited time to listen to and speak with their patients.

What about genetics online?

These commercial sites provide information on basic genetic principles, on mechanisms of inheritance, on diagnostic criteria and manifestations of common genetic disorders. They provide links to support group organizations. But I found a striking lack of links to diagnostic laboratory and professional genetic services.

The www.Healthfinder.gov site maintained by the NIH. When searched for genetics it provided many useful links, most of them to other government sites including PubMed and OMIM.

Here are some of the genetics resources we all know and use.
GeneClinics and the associated GeneTests, formerly Helix, supported by NIH and directed by R. Pagon at the Univ. Wash provides well-organized profiles of genetic diseases with emphasis on diagnosis and molecular testing. Designed primarily for healthcare providers the sites are now accessible also to consumers. [http://www.geneclinics.org/index.html]

This is only part of what is available and it is only the beginning.

There is a huge need to provide online genetic information tailored to consumers that is accurate, up-to-date and accessible. There should be career opportunities for site developers and data base curators.

The availability of health-related web information to the general public has a significant effect on our practice of medical genetics and genetic counseling. I don't believe this is unique to our local geography, but, practicing within Silicon Valley, we may see more of it than other parts of the country.

Patients will walk in with a stack of print-outs from the web including the latest article on treatment for their disorder that you may not have seen yet - or with papers you have written - and they have very specific questions. Lacking formal training in biomedicine, their understanding may be incomplete and the possibilities for misunderstanding are great.

An extreme case is a client who has diagnosed herself with a genetic disease, based on extensive web searches, is looking for a physician to confirm it and cannot be convinced otherwise. We all know the medical student syndrome, as you learn about diseases you discover symptoms in yourself; medical news writers can suffer from this too, I'm told.

In other cases, web searches by patients or their relatives did in fact lead to the correct diagnosis that had been missed by healthcare professionals. More and more, we are dealing with highly informed clients. That changes the nature of the clinical encounter and the content of the counseling we provide.

Will medical genetics services be provided online? On an individualized basis? Frankly, I cannot think of any medical specialty better suited for such a development. Exchange of information is the major part of our practice. Medical genetics is a cognitive, not a procedure-oriented specialty.

As a professional society, we have long been concerned about the discrepancy between the
explosion of new knowledge and the shortage of trained clinical geneticists and counselors who can convey this information and provide services on a one-on-one basis. It seemed like an impossible task to accomplish. The internet offers a solution. Web-based genetic information will increase the awareness of genetic conditions and of the role of genetic factors in common diseases among the public. Awareness creates more requests for services. I have no doubt that interactive web-based systems will be developed that can provide accurate, timely and individualized genetic information.

When I was a child, we had no TV, no telephone, no central heating and no water-flushing toilet - we had a radio, newspapers and books. Momentous changes have taken place within our lifetime.

At the threshold of a new millennium, the information age is here. There is no turning back. This revolution is happening now. We are a very special generation in human history. It is a privilege to be alive today.

For those of you currently in training, this revolution offers unprecedented opportunities for new career paths. For us more advanced geneticists, this revolution offers unprecedented opportunities to adapt our professional roles for maximal impact in this new environment. It is up to each of us individually, and as a Human Genetics Society, to be creative and take up the challenge.

References


