

INNOVATIONS IN HUMAN GENETICS EDUCATION

Using Medical Genetics Applications to Educate for Computer Competence

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Summary

This article proposes specific areas of computing competence and illustrates how these skills can be acquired as an integral part of the curriculum of medical genetics. Geneticists are at the forefront in the use of computers for medical care, because of the driving force of the Human Genome Project. Computer searching of international data bases is the most efficient method to keep current with the explosion in molecular genetics data and with its immediate relevance to clinical care. The use of computers in genetics education could go far beyond the use of computer-assisted instruction (CAI) to show how to use computer systems to assist with clinical decisions. The proposed basic computer skills can be obtained using genetics software. The six proposed skills include the use of (1) microcomputers, (2) productivity software, (3) CAI, patient simulations and specific application programs, (4) remote computers, (5) data bases and knowledge bases, and (6) computers to improve the clinical care of patients.

Introduction

The Human Genome Project and the increasing availability of international data bases mandate advanced computer knowledge as a prerequisite to the practice of medical genetics. Students, fellows, and genetic counseling students need to know how to access and use this information as part of quality medical care of patients, and it is a priority of many academic medical centers.

The majority of medical colleges accept the concept of basic computer competence being necessary for the practice of medicine (Pages et al. 1983; AAMC 1984; Starkweather 1986; Salamon et al. 1989), but many colleges are still looking for model programs which

demonstrate the actual use of computers to assist with clinical decisions and patient care. The present article proposes specific areas of computing competence and illustrates how these skills may be acquired as part of a model curriculum of medical genetics. The use of computers in genetics education could extend beyond traditional computer-assisted instruction (CAI) to the use of current genetics data immediately available with computer technology.

The Proposed Basic Computer Competencies

The proposed basic computer skills needed to practice medicine include the following:

- I. Basic microcomputer literacy
- II. The use of productivity software
- III. The use of CAI, patient simulations, and specific application programs
- IV. The access and use of remote computers
- V. The understanding and utilization of data bases and knowledge bases.

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VI. The use of a computers to improve clinical care.

The boundaries between these six areas are somewhat indistinct. In the day-to-day practice of medicine, the six areas are used concurrently to improve problem-solving skills, hone thought processes, and keep current with recent developments. This is essentially the definition of competency VI, the ultimate goal of this learning.

I. Basic Microcomputer Literacy

Today's computer literacy focuses on desktop microcomputers and not on mainframe computing. A maximum of 2–3 h is needed to acquire basic computer literacy. The basic skills include how to turn on the computer, how to take care of floppy disks, basic file management capabilities, how to interact with the computer, and the use of either special keys on the keyboard or devices such as a mouse. All students should have these skills before entering medical school.

II. The Use of Productivity Software

Students need to know enough about productivity software to understand the different functions of software products, to recognize when they need to be employed, and to be able to become competent in their use. The basic types of productivity software include word processing, electronic mail, graphics, spreadsheets, and data bases.

There are several methods by which students could be working on genetics projects using productivity software. The educator could (a) give a writing assignment and require that it be done on a *word processor*; (b) have students submit their assignments via *electronic mail*; (c) have the students consider either gene frequency change over several generations or a laboratory-based data analysis and require them to use a *graphics package* to display their findings; (d) have the students use a *spreadsheet* to calculate changes in gene frequency, risks, and Bayesian conditional probabilities and to store laboratory experimental results; and (e) use a *data base* to keep a concise record of all patients they see (Oliver et al. 1990), all procedures they do, the concepts and diseases they learn, the data they collect on a specific experiment, and the literature references they read. For fellows and trainees, these data bases would be particularly useful for preparation of material for national board examinations.

III. The Use of CAI, Patient Simulations, and Specific Application Programs

Any list of these programs is out of date as soon as it is published, since this is a very active area of software development; the intent of the present article is not to list every conceivable software product but to illustrate this overview with some specific examples. The essential skills for the students include (1) understanding what is taught by each package, (2) using the program on one's own, and (3) recognizing its utility for lifelong, independent learning. Students should be knowledgeable appraisers of new software, capable of questioning the value of a new package while determining the equipment needs to make it operational.

In the arena of multimedia and interactive videodisc programs there are many application packages being developed in all categories of CAI. The true power of multimedia involves learning by multiple senses and multiple computer modalities. Developments in the areas of graphics overlays, CD-ROM for large storage capacity, videodiscs for pictures and motion, audio cards for speech and sound synthesis, and a hypertext arrangement of educational material promise to transform CAI into a more effective experience.

A. CAI tutorials. — One tutorial package, Genetics Applications, is a 10-disk package to teach the basic principles of medical genetics (see Appendix for addresses of sources of all software discussed). Institutions use it as background material for their students, as a complete course, and as a review mechanism for trainees wishing to prepare for national board examinations. A more basic package is called Introductory Genetics.

B. CAI drill and practice. — Drill and practice programs deliver multiple-choice questions via the computer, augmented with assistance in those cases when the student makes an incorrect choice of answer. Some programs are strictly for practice, and some programs will deliver the actual test, grade it according to the faculty key, and run an item analysis of the class results.

The Medical Genetics Question Bank (Reed et al. 1989) is an example of a well-developed question bank for medical genetics. It could be used in almost any genetics course for review of material prior to an examination and has adequate but concise explanations of the incorrect answers to guide the students in further learning. MacGeneRisk is a computer program which administers Bayesian risk-estimation problems with blanks in sentences, to be replaced by words or numbers.



It is probably inevitable that national examinations will be administered via a computer, so students should become comfortable with the computer as a testing agent. Multiple-choice problems and patient-management simulations are the most likely format for the initial round of national computer-administered examinations.

C. Patient simulations. — Computerized patient simulations provide students the opportunity to practice diagnosis and management skills on simulated patients rather than on real patients. The goal is not to replace real patient experience by the computer but to augment the educational experiences and allow the student to capitalize on the real patients seen. These programs help students to develop a specific deductive reasoning approach to medical problem solving and to incorporate it into the process of patient care.

In genetics, the available simulation programs are Galactosemia, Down Syndrome, and Fragile X Syndrome. These programs concentrate more on management of specific complications than on diagnosis but are still useful for learning general management principles and follow-up protocols.

D. Specific application programs. — Many specific genetics programs could be used in genetics education. One program already widely used is Possum (Picture Of Standard Syndromes and Undiagnosed Malformations), a data base about syndromes and a videodisc library of pictures pertaining to the syndromes. The main use of the system is to assist in diagnosis (Strmme 1990), but many geneticists use the system when teaching dysmorphology and encourage “browsing” the picture archive.

Most application programs are built to do a specific task: clinic management (GOAS, or GenetiWare), patient registries (Pratt and Mize 1990), risk estimation, research-related calculations, pedigree drawing (Megadats or Pedigree/Draw), chromosome analysis, mapping (MicroMap), height prediction (Advanced Height Prediction), or sequence-similarity searching (Altschul et al. 1990; Altschul and Lipman 1990; Karlin and Altschul 1990), etc. These programs could be used in an educational mode to acquaint students with the programs and their appropriate uses. For example, the educator could use a local data-base management system such as GOAS (Cutts et al. 1988) to teach about the principles of data bases and to give students specific exercises to allow them to become familiar with searching the data base to answer patient-care or research questions.

IV. The Access and Use of Remote Computers

Students need to learn to access patient data stored in a hospital mainframe system, references in literature data bases, bulletin boards of specialty societies, remote data bases with information useful for medical practice, and international data networks. Students should learn about national telecommunications networks and their methods of access. Specifically, students should learn to (a) use a modem, (b) obtain network access to the Internet, (c) log-on to a remote computer system, (d) upload and download files, and (e) operate the remote system once on-line.

To assist students in learning these skills, an educator could pick a particular system of interest, demonstrate the access procedures and use, discuss the modem settings or network protocols, demonstrate the methods to upload and download data, and show an actual use of the system. The students could solve a similar problem by using the remote system as a tool. One example of this methodology for genetics education (Proud et al. 1989, and submitted; Mitchell et al. 1990, and in press) uses Grateful Med for MEDLINE access. Besides MEDLINE, other remote systems include COMPUSERVE to post a patient-care question on the medical form, BITNET to send a message to a colleague, OMIM to search for information about Mendelian conditions, TERIS or Reprotox to find reproductive toxicology data on drugs/substances in pregnancy, and NFormation to access an on-line bulletin board focused on neurofibromatosis.

V. The Understanding of and Utilization of Data Bases and Knowledge Bases

The need for scientific research data places genetics in a rather unique situation. The research results are being collected and made available to the research community in nationally maintained data bases. Those results are available electronically months or years before they are available in the published literature. Much information about many specific genetic diseases that is in textbook form is out-of-date before the textbook is published. Even the journal literature may be out-of-date before it is published. The most accurate and current data and their derived knowledge will increasingly be in electronic format.

Unfortunately, the information is only available to those who understand the basic structure of a data base and knowledge base and the principles of their use so that retrieval is accurate and complete. A *data base* stores basic data in a structured format (fields,



records, relationships between data bases, and specific vocabulary for field entries) and can be queried to determine which set of data matches a specific question. A *knowledge base* stores knowledge in a structured format (inference rules, decision trees, knowledge frames, and semantic networks) and can be utilized to determine whether sets of data match predetermined patterns, diagnoses, or conditions. For example, a dysmorphology-patient data base would store features of syndromes. A knowledge base might store the rules to search the patient data to determine which patients would either be candidates for a specific treatment modality or have a specific diagnosis.

Students need adequate training in the major literature data bases (MEDLARS) and in the MeSH (*Medical Subject Headings*) vocabulary to index the articles used. Teaching about other data bases would include a discussion of the format and vocabulary needed for accurate retrieval. Most national data resources will accommodate special teaching projects and assist with student access to these various systems.

Knowledge-base examples are rather more unusual in genetics but promise to become more common over the next few years. Both the Birth Defects Information System (Buyse and Edwards 1987) and Syndroc (Schorderet 1987) use a knowledge base to make diagnostic suggestions. In the field of molecular biology, the systems built to assist with planning of biotechnology experiments (Brutlag 1990) promise to be useful for basic scientists. The ISCN expert system (Cooper and Friedman 1990) interprets the International Standard Chromosome Nomenclature and outputs the actual chromosome anomaly. The AI/GEN Deaf-Blind expert system (Mitchell et al. 1985) achieved results in helping to define diagnostic criteria. Other systems are currently being developed (Patton 1987; Baraitser et al. 1989; Veloso and Feijoo 1989; Salgado et al. 1990; Yamamoto et al. 1990).

VI. The Use of Computers to Improve Clinical Care

The ultimate goal is to use a computer to improve clinical care. Computers can assist in all aspects of developing problem-solving skills, honing thought processes, and keeping current with the latest information for clinical care. Consider the following two realistic examples of using currently available systems:

1. Dr. Mary Smith, a medical geneticist, is caring for a patient with neurofibromatosis type 1 (NF1) and wishes to advise the patient and family on the latest information on carrier testing, prenatal diagnosis, treatment, and risks to other family members.

Dr. Smith would log-on to the GDB (Genome Data

Base) and OMIM (*Online Mendelian Inheritance in Man*) (either by modem or via the Internet) to ask (a) what is the status of mapping, cloning, and sequencing the gene for NF1, and (b) what is the current status of carrier testing, prenatal diagnosis, and treatment for NF1? She would see a discussion of the molecular biology studies both on the gene located at 17q11.2 and on the flanking markers and would note that the information had been updated within the past 10 d. She connects to The American Society of Human Genetics Bulletin Board (discontinued July 1, 1991) to search the abstracts of articles which have been accepted for publication but which are not yet in print; she finds that, within the next 6 mo, 12 articles discussing sequencing, mapping, molecular diagnosis, and prenatal diagnosis of NF1 will be published. Dr. Smith could delve further into the actual DNA and protein sequences by using GENBANK and the PIR (Protein Information Resource) with sequence-similarity search algorithms (Altschul et al. 1990; Altschul and Lipman 1990; Karlin and Altschul 1990) but decides to concentrate on the patient tasks at hand and to leave molecular details to the follow-up visits. Dr. Smith calls the lead author of one of the abstracts to discuss prenatal diagnosis of NF1 and to determine whether the family she is treating would fit into their abstract's experimental protocol. Finally, after appropriate testing but before giving the patient and family the recurrence risks, Dr. Smith could use her spreadsheet and calculator to determine the risks to her patients. Of course, Dr. Smith would want to search her local genetics data base (a) to determine whether there are other NF1 families who are living near the patient and who would provide emotional support if needed or (b) to see whether another branch of this family had been seen previously in her center. Dr. Smith could accomplish all of these activities from the personal computer sitting on her desk. The actual computer time would probably be less than ½ h.

2. Dr. John Baker, a medical geneticist, is asked to consult on a case of a child with multiple dysmorphic features whose mother took drug X in pregnancy.

After examining the child, Dr. Baker connects to the ReproTox system to see whether drug X had been implicated in specific birth defects; he finds a retrospective study of the drug which has rather nonspecific malformations associated with first-trimester ingestion. He connects to the locally mounted London Dysmorphology Data Base to assist with a differential diagnosis by the key dysmorphic features. He finds three conditions which seem to have similarities to those of his patient, gets a list of appropriate reprints,



and requests them electronically from his library; they are delivered within the hour. He focuses on one rare syndrome which he has not seen previously. He checks the POSSUM system to consider both his patient's similarities to previous cases of the rare syndrome and other potential diagnoses. Dr. Baker does a MEDLINE search using Grateful Med and also checks, via The American Society of Human Genetics Bulletin Board (discontinued July 1, 1991), the abstracts of articles accepted for publication in the *American Journal of Medical Genetics*; he finds a recent case report of the rare syndrome which was associated with maternal ingestion of drug X. A colleague at the Karolynska Institute in Stockholm wrote the report. Dr. Baker sends his colleague an electronic-mail message to discuss the case; the colleague has a reply by the next morning and updates Dr. Baker on the case-management details of his patient. Dr. Baker puts patient photographs in the mail to his colleague in order to confirm the diagnosis. He calls to request a check of the Great Plains Genetics Services Network data bases to determine whether any other patients with this disease have been reported in that eight-state region; there was one reported case in Missouri, so Dr. Baker calls to discuss the case with the geneticist. Dr. Baker then checks his local genetics data base to determine whether his center had seen any other patients with a similar constellation of symptoms or who were prenatally exposed to drug X; after checking the records of the three patients who might seem similar, he decides that his patient is unique but that two of the other patients may need a reevaluation (he assigns follow-up of the two additional patients to his research fellow). Dr. Baker has extensively used electronic information to assist with this consult; he has decided on the most probable diagnosis, consulted with colleagues both nationally and internationally, developed a plan for confirming the diagnosis, and instituted a management plan.

In summary, these scenarios show efficient problem solving using a variety of electronic information sources to improve clinical care. It is an augmented "invisible college" (Perry and McKinin 1990), since, although phone calls to colleagues are still important steps, it recognizes that much important data need not be delayed by busy professionals who are not easily reached by telephone.

How Do Geneticists Investigate these Educational Models?

For those genetics educators who wish to explore these suggestions in more detail, there are several ave-

nues available at most academic institutions to assist with incorporating computer use into the curriculum. An educator could (1) contact the computer center for short courses in specific software packages, especially the productivity software, and ask that students take the course as a prerequisite; (2) plan to attend a short course held by a commercial vendor to assist with the optimal use of the software; (3) discuss the planned computer intervention with the unit responsible for computers in the curriculum; (4) inquire about individual tutoring or assistance with a specific program for students who need special assistance; (5) ask the medical or university librarians about assistance in teaching access methods and in searching of national data bases.

When using computer programs in the curriculum, the educator can discuss the different techniques involved in the computer applications being used. For example,

1. when using a program on Advanced Height Prediction, emphasize that the results are based on three standard algorithms for estimating height based on the input variables. It is useful to teach the algorithms and the underlying principles, but it is equally important to recognize the validity of a computer calculation and the ease of use of the program.
2. when using the London Dysmorphology Data Base (Boda et al. 1988-89), teach the methods of searching the data base with standard Boolean logic. When students encounter another data base, they will know the basic terminology and principles of searching data bases.
3. when recommending a patient simulation in Galactosemia, discuss the decision branchpoints of the program, the methods of the simulation (probabilistic, set branching, or hypertext), and how the structure of a patient simulation is standard with recognizable parts. Students will have a better background for the next simulation.

These suggestions are only the beginning of the education of students in the uses of computers in medicine. Medical genetics has always been the leading force in the use of quantitative information; it could be the leading force in computer usage by the medical profession as well.

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Appendix

Contact Information for the Software Discussed

Advanced Height Prediction

Arc Software
23131 Cumorah Crest
Woodland Hills, CA 91364

BITNET or Internet

Check with the local computer center or use COM-
PUSERVE for access.

COMPUSERVE

Corporate Headquarters
5000 Arlington Centre Boulevard
P. O. Box 20212
Columbus, OH 43220
Phone (800) 848-8990

Galactosemia, Fragile X, and Down Syndrome Health

Sciences Consortium
201 Silver Cedar Court
Chapel Hill, NC 27514
Phone (919) 942-8731

GENBANK

To submit DNA sequencing to GENBANK, phone
(505) 665-2177
For data base distribution information, contact
IntelliGenetics
700 East El Camino Real
Mountain View, CA 94040
Phone (415) 962-7300
FAX (415) 962-7302

Genetics Applications

Learner Managed Designs, Inc.
2201 West 25th Street, Suite K
Lawrence, KS 66046

Genetics Office Automation System (GOAS)

Contact Joyce A. Mitchell, Ph.D.
605 Lewis Hall
Medical Informatics Group
University of Missouri
Columbia, MO 65211
Phone (314) 882-6966

GentiWare

MediSoft Creations
Contact Frank Greenberg, M.D.
Institute of Molecular Genetics
Baylor College of Medicine
One Baylor Plaza
Houston, TX 77030

Great Plains Genetics Services Network Data Base

Contact James Smith
Medical Genetics Division
University of Iowa
Iowa City, IA 52242
Phone (319) 353-6687

Grateful Med version 5.0 (MEDLINE access system)

U.S. Department of Health and Human Services
National Library of Medicine
MEDLARS Management Section
8600 Rockville Pike
Bethesda, MD 20894
Phone (800) 638-8480; in Maryland
(301) 496-6193

Introductory Genetics

Educational Materials and Equipment Company
Old Mill Plain Road
Danbury, CT 06811

London Dysmorphology Data Base

Oxford Electronic Publishing
200 Madison Avenue
New York, NY 10016
Phone (212) 679-7300

MacGeneRisk 1.04

Contact John H. Caster, Ph.D.
Department of Medical Education
School of Medicine
Southern Illinois University
Carbondale, IL 62901
Phone (618) 536-5513

MEGADATS

Helios Software Works
P.O. Box 40068
Indianapolis, IN 46240-0068
Phone (317) 253-6965

NFormation

The Neurofibromatosis Institute
11 West Del Mar Blvd.
Pasadena, CA 91105
Phone (818) 356-3400
Log-on phone (818) 957-3508

OMIM and GDB

Laboratory for Applied Research in Academic
Information
William H. Welch Medical Library
The Johns Hopkins University
1830 East Monument Street, Third Floor
Baltimore, MD 21205



Pedigree/Draw for the Apple Macintosh, Version 3.0
 Contact P.M. Mamelka, B. Dyke, or J. S. MacCluer
 Department of Genetics
 SouthWest Foundation for Biomedical Research
 P.O. Box 28147
 West Loop 410 at Military Dr.
 San Antonio, TX 78284
 Phone (512) 674-1410

PIR (Protein Identification Resource)
 National Biomedical Research Foundation
 Georgetown University Medical Center
 3900 Reservoir Road, N.W.
 Washington, DC 20007
 e-mail PIRSUB@GUNBRF.BITNET

P.O.S.S.U.M. Project
 C.P. Export Pty. Ltd.
 613 St. Kilda Road, Melbourne
 Victoria 3004 Australia
 Phone 613-520-5333

ReproTox (Reproductive Toxicology Information System)
 Reproductive Toxicology Center
 2425 L Street, N.W.
 Washington, DC 20037-1485
 Log-on phone (800)535-4100

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- posium. School of Health Information Science, University of Victoria, Victoria, British Columbia
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