Understanding Among Health Professionals

Direct-To-Consumer Genetic Testing

National Academy of Sciences

1 September 2009

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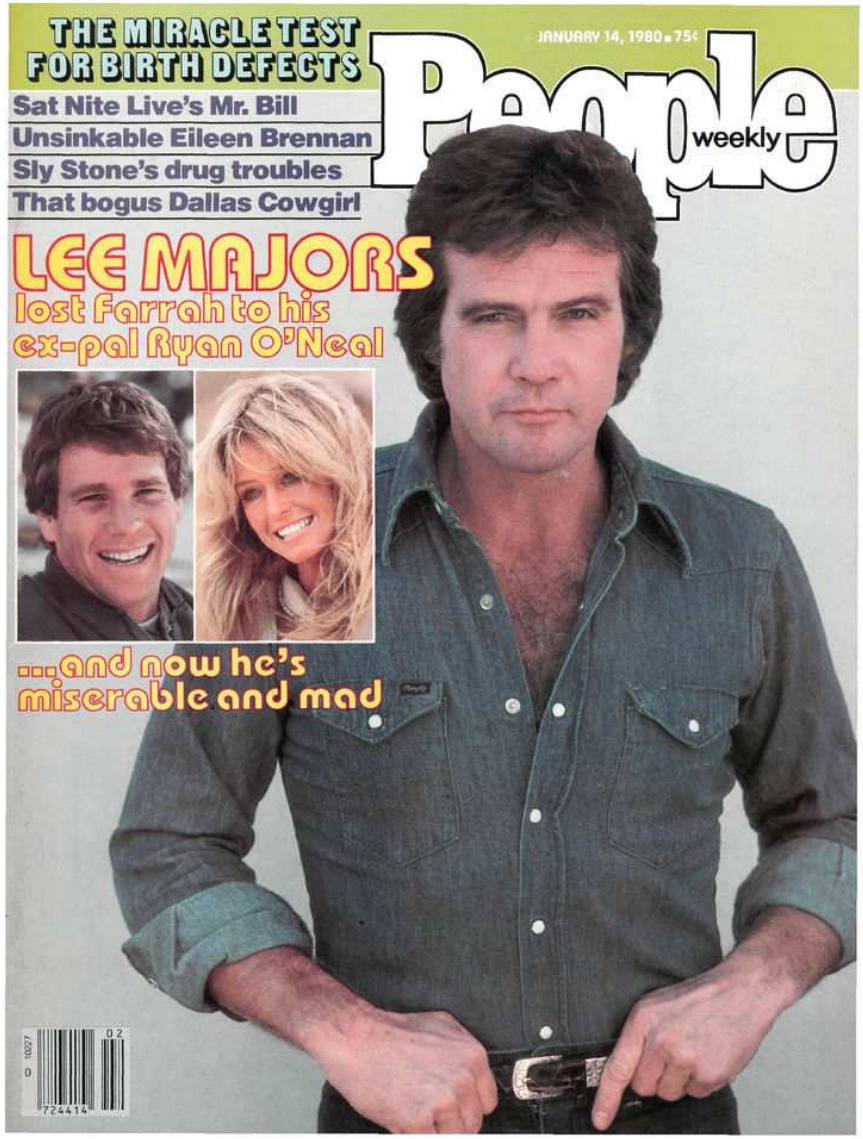
NCHPEG
National Coalition for Health Professional Education in Genetics

“Many issues raised in relation to DTC services reflect larger issues in the health-care system.”

Elissa Levin - Navigenics

I hope to accomplish the following:

- Review some information about health professionals' understanding of genetics
- Review some challenges and opportunities with respect to educating health professionals about genetically based health care
- Comment on the relationship between public education and the education of health professionals
- Distinguish personalized medicine from individualized medicine



January 14, 1980 Vol. 13 No. 2

U.S. System of Oversight of Genetic Testing:

Secretary's Advisory Committee on Genetics, Health, and Society
April 2008

"(Practitioners) cannot keep up with the pace of genetic tests (and) are not adequately prepared to use test information to treat patients appropriately….Practice guidelines are insufficient to ensure appropriate care."

REVIEW CLINICIAN'S CORNER

Delivery of Genomic Medicine for Common Chronic Adult Diseases

A Systematic Review

Maren T. Scheuner, MD, MPH
Pauline Sieverding, MPA, JD, PhD
Paul G. Shekelle, MD, PhD

"…the primary care workforce, which will be required to be on the frontlines of the integration of genomics into the regular practice of medicine, feels woefully unprepared to do so."

Wolters Kluwer Health | Lippincott Williams & Wilkins

ACADEMIC MEDICINE
Journal of the Association of American Medical Colleges

Preceded by: Journal of Medical Education (ISSN: 0022-2577)

Association of American Medical Colleges
Volume 82(5), May 2007, pp 441-445

The Current Status of Medical Genetics Instruction in U.S. and Canadian Medical Schools

Thurston, Virginia Carol PhD; Wales, Paula Sue EdD; Bell, Mary Alice MS; Torbeck, Laura PhD; Brokaw, James Joseph PhD, MPH

Data from the paper by Thurston et al.

- 149 U.S. and Canadian course directors in medical genetics or curricular deans in accredited medical schools
- Response rate = 75.2%, as of June 2005
- 77% = medical genetics in 1st year
- 47% = incorporated into 3rd or 4th year

Data from the paper by Thurston et al. (cont'd.)

- 62% = 20-40 hrs of instruction
  - 86% = general concepts
  - 11% = practical applications
- 46% = stand-alone course
- 54% = integrate med genet into another course

Wolters Kluwer Health | Lippincott Williams & Wilkins

Genetics IN Medicine
Official Journal of the American College of Medical Genetics

Providers' knowledge of genetics: A survey of 5915 individuals and families with genetic conditions

Harvey, Erin K. ScM, CGC; Fogel, Chana E. MGC; Peyrot, Mark PhD; Christensen, Kurt D. MPH; Terry, Sharon F. MA; McInerney, Joseph D. MA, MS

Volume 9(5), May 2007, pp 259-267

Challenges to Genetics Education for Health Professionals (not limited to physicians)

- Crowded curriculum
- Misconceptions about genetics
- Lack of knowledgeable faculty
- Disconnect between basic sciences and clinical experiences during training
- Failure to integrate genetics across the curriculum (two efforts underway)
- Inadequate representation of genetics on certifying exams

Guttmacher AE, Porteous M, McInerney JD. *Nat Rev Genet,* Feb 2007

Some Challenges to the Integration of Genetics into Primary Care

- Dearth of genetics professionals*
- Lack of knowledge about genetics among primary-care providers*
- Lack of confidence *
- Lack of referral guidelines *
- Difficulty interpreting genetic tests
- Difficulty explaining genetic risks to patients

*Suther, S. and Goodson, P. Barriers to the provision of genetic services by primary care physicians: A systematic review of the literature. *Genet Med* 5(2): 70-76, 2003.

"Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence.

- Education about inheritance, testing, management, prevention, resources, and research.

- Counseling to promote informed choices and adaptation to the risk or condition."

Resta R, Biesecker BB, Bennett RL, et al. 2006. A new definition of genetic counseling: National Society of Genetic Counselors' task force report. *Journal of Genetic Counseling* 15(2): 77-83.

Genetics is a cognitive discipline, and genetic information has intrinsic value, but

"Thinking is not highly valued in the healthcare reimbursement system."

Epstein CJ. 2004. Genetic testing: Hope or hype. *Genetics in Medicine* 6(4): 165-172.

Central Questions

- Which content is appropriate?
  - Accurate vs. complete
- Who needs to know what?
- Which clinical behaviors and attitudes do we want to change, and can we?
- How do we define and measure success?
- How do we deliver instruction most effectively, e.g., CME, EMRs, grand rounds; point of care

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Genetics IN Medicine

Official Journal of the American College of Medical Genetics

September 2008,Vol. 10 No. 9

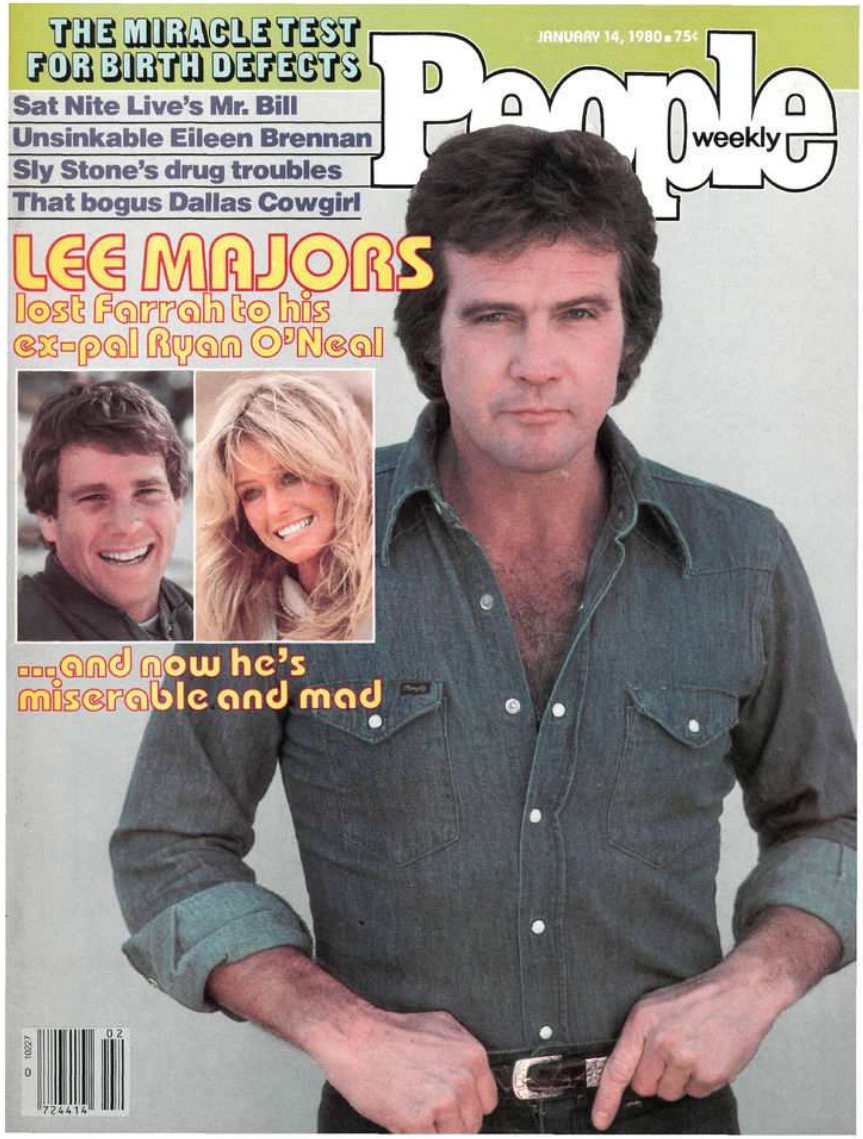
Twenty questions in genetic medicine—an assessment of World Wide Web databases for genetics information at the point of care

Howard P. Levy, MD, PhD, Leigh LoPresti, MD, and Diane C. Seibert, PhD, CRNP

"Many World Wide Web databases do not answer clinical questions about genetic conditions accurately. ***None of the resources we tested are efficient enough for point-of-care use.*** *As genetics becomes more prominent in daily patient care, providers will need an efficient, accurate, and accessible source of information."*

A Distinction

- Personalized medicine: A way of practicing that is rooted in tests, technologies, and procedures (and that is not limited to genetic medicine)
- Individualized medicine: A way of thinking that is rooted in genetics (the study of inherited biological variation) and evolution (the impact of variation on adaptive and disadaptive phenotypes in the context of the environment) – a counterpoint to typological thinking



January 14, 1980 Vol. 13 No. 2

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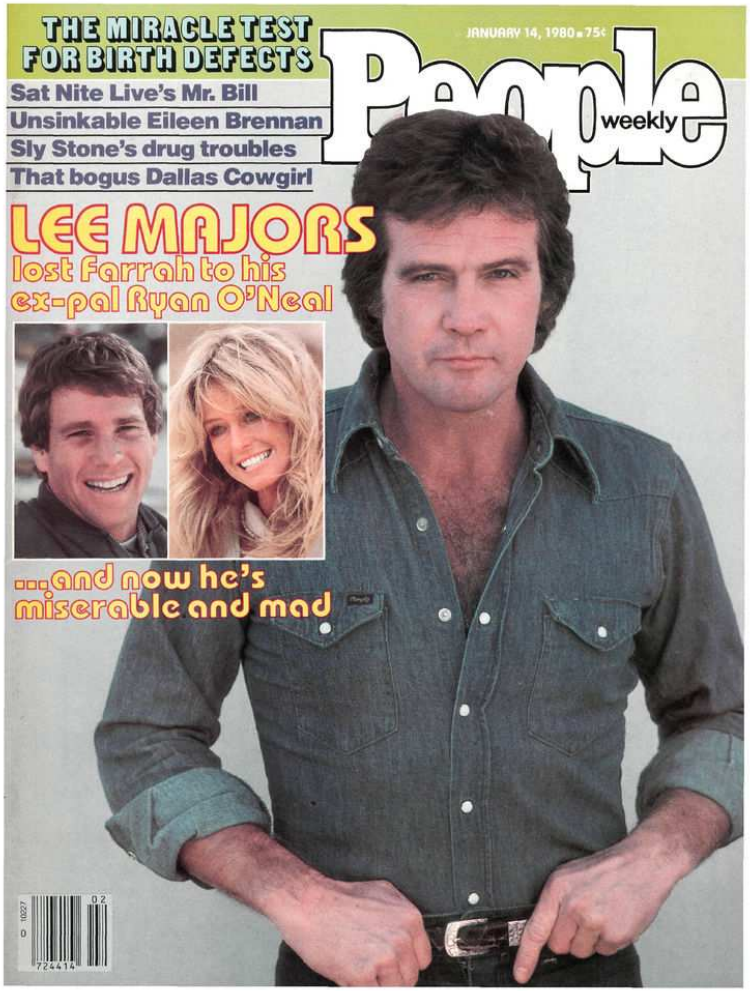
Pregnancy After 35 Has Risks, but Amniocentesis Can Alert Mothers in Time, Says An Expert

By Gail Jennes

As more women postpone childbearing until their mid-30s, often for career reasons, a new element of risk is added: Mothers of 35 and older give birth to nearly one-fourth of all babies with mongolism, although such women constitute only about five percent of all mothers.



Genetic Literacy for the Public

The knowledge and skills the average person needs to manage uncertainty and to participate as a full partner in a prevention-based health-care system that increasingly is informed by genetic perspectives

Education for the public and health professionals should be complementary.

Early Insights into DTC Testing

Characteristics of users of online personalized genomic risk assessment: Implications for physician-patient interactions. McBride CM, et al. August 2009.

- Survey participants did not "hold genetically deterministic explanations for common preventable health conditions."

- "Participants reported high levels of confidence in their ability to navigate the health-care system…and to understand genetics."

the spittoon
more than you've come to expectorate

Curriculum reform needed for an informed public when it comes to genetics, expert says

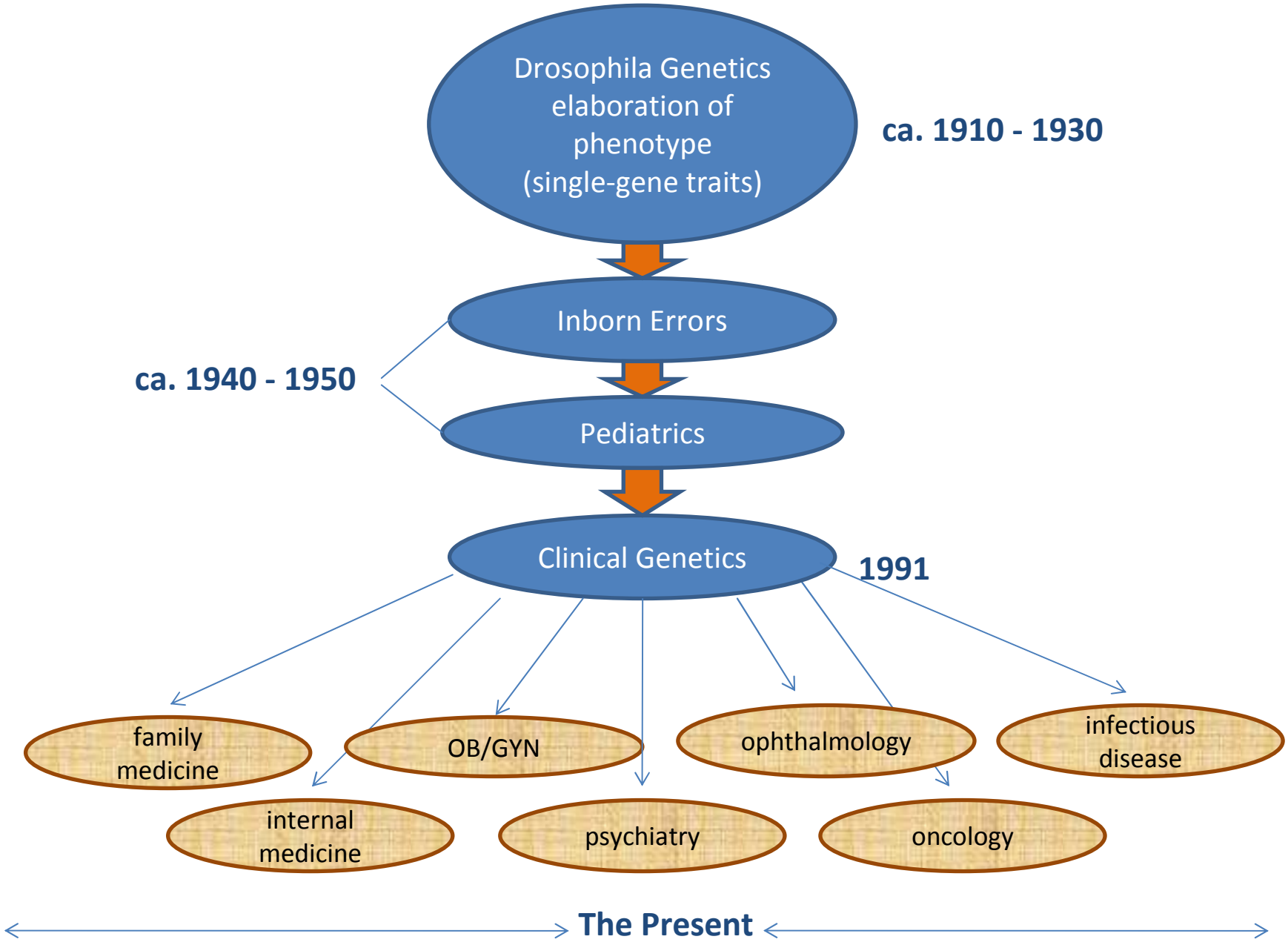
"It's not enough to teach genetics, says Michael Dougherty, director of education for the American Society for Human Genetics. It has to be taught in the right way." (25 June 2009)

Closing the Gap: Inverting the Genetics Curriculum to Ensure an Informed Public

Michael J. Dougherty

Am. J. Hum. Genet. 85(1): 6-12, 2009

"…the predominant mode of genetics instruction (a focus on Mendelian traits) primes many students to think deterministically and with a confused understanding of risk….It might be preferable to…begin genetics instruction with common quantitative traits, which might include health and disease traits but should not be limited to them…."


Drosophila Genetics
elaboration of
phenotype
(single-gene traits)
ca. 1910 - 1930
Inborn Errors
ca. 1940 - 1950
Pediatrics
Clinical Genetics
1991
family
medicine
OB/GYN
ophthalmology
infectious
disease
internal
medicine
psychiatry
oncology
The Present

This might be the first time in the history of organized medicine that we are trying to decentralize – informally – a certified medical specialty so it pervades all of the other specialties.

A Modest Proposal To Help Integrate Genetics into Education and Mainstream Health Care

Stop using the terms “genetic disorder” and “genetic disease.”

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