Genomics in health related education

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Quality of Care

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The Human Genome Project:

- # genes 20,000-25,000


### OMIM Statistics for November 21, 2008

#### Number of Entries

<table>
<thead>
<tr>
<th>Category</th>
<th>Autosomal</th>
<th>X-Linked</th>
<th>Y-Linked</th>
<th>Mitochondrial</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>* Gene with known sequence</td>
<td>11886</td>
<td>568</td>
<td>48</td>
<td>37</td>
<td>12539</td>
</tr>
<tr>
<td>+ Gene with known sequence and phenotype</td>
<td>348</td>
<td>27</td>
<td>0</td>
<td>0</td>
<td>375</td>
</tr>
<tr>
<td># Phenotype description, molecular basis known</td>
<td>2202</td>
<td>204</td>
<td>2</td>
<td>26</td>
<td>2434</td>
</tr>
<tr>
<td>% Mendelian phenotype or locus, molecular basis unknown</td>
<td>1525</td>
<td>138</td>
<td>5</td>
<td>0</td>
<td>1668</td>
</tr>
<tr>
<td>Other, mainly phenotypes with suspected mendelian basis</td>
<td>1929</td>
<td>140</td>
<td>2</td>
<td>0</td>
<td>2071</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>17890</td>
<td>1077</td>
<td>57</td>
<td>63</td>
<td>19087</td>
</tr>
</tbody>
</table>

#### Synopsis of the Human Gene Map

<table>
<thead>
<tr>
<th>Chr.</th>
<th>Loci</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

2007

11941
379
2221
1595
2107
18243
Genetics & health

- The greatest public health benefit of advances in understanding the human genome may be realized for common chronic diseases such as cardiovascular disease, diabetes mellitus, and cancer (Scheuner 2008).

- Physicians lack oversight of genetic testing and concerns about privacy and discrimination, and report inadequacy to deliver genetic services (Scheuner 2008).

- Physicians lack knowledge of genetics relevant for daily practice (Baars et al. 2005).
Passing scores according to nongenetic health care providers:

- Essential: 85%
- Desirable: 65%
- Too specialized: 48%

Conclusions: Medical students nearing graduation lack genetic knowledge that is essential for daily practice. Therefore, changes should be made in the medical curricula. *Genet Med* 2005:7(5):295–301.
Genetics & health

- For genomics to have an effect on clinical practice that is comparable to its impact on research will require advances in the genomic literacy of health-care providers (Guttmacher 2007).

- Genetics of common disorders (Julian-Reynier et al, 2008)

• Paper Boerwinkel et al.

- Major changes are apparent...

- What changes has genomics brought to society, esp. Medicine

• FEW!!!

• If existing diagnostic/therapeutic channels fit: easier than prevention

Mammaprint®
Genetics & health

- Paper Boerwinkel et al.
  - From single genes to genome wide studies
  - From monogenic disorders to common disorders
Genetics & health

• Paper Boerwinkel et al.

• Q1: methods changed? GWAS, hypothesis free, valorisation of tests for susceptibility without clinical utility -> protection of consumers needed

• Q2: in medical curriculum: when discussing causes of common disease, include genetic susceptibility

• Q3: Direct to consumer testing; 1000$ genome; consumer genomics

• Q4: Discuss pros and cons of potential screening test, understand “clinical utility”, prepare for life-long-learning

• Q5: some are..... Hypothesis free; DTC

• Q6: but they don’t think about >1Y
CANMEDS: the doctor in patients’ eyes

Medical Expert
Communicator
Collaborator
Manager
Health Advocate
Scholar
Professional

Reflector
COMMUNICATOR
- discuss appropriate information the health care team

COLLABORATOR
- consult effectively with other physicians and health care professionals
- contribute effectively to other interdisciplinary team activities

MANAGER
- utilize information technology to optimize patient care, life-long learning, other activities

HEALTH ADVOCATE
- identify the important determinants of health affecting patients
- contribute effectively to improved health of patients and communities
- recognize and respond to those issues where advocacy is appropriate

Scholar
- develop, implement and monitor a personal continuing education strategy
- critically appraise sources of medical information

Professional
- deliver highest quality care with integrity, honesty and compassion
How to build a curriculum? (Q9-10)

• Problem based, patient central, core competencies not defined for genetics/genomics (or anatomy or physiology)
Perspectives

Science and Society

Educating health-care professionals about genetics and genomics

Alan E. Guttmacher, Mary E. Porteous and Joseph D. McInerney

Abstract | To biomedical researchers, this is the ‘genome era’. Advances in genetics and genomics such as the sequence of the human genome, the human haplotype map, open access databases, cheaper genotyping and chemical genomics have already transformed basic and translational biomedical research. However, for most clinicians, the genome era has not yet arrived. For genomics to have an effect on clinical practice that is comparable to its impact on research will require advances in the genomic literacy of health-care providers. Here we describe the knowledge, skills and attitudes that genomic medicine will require, and approaches with unique biology, not some biological everyman, which limits the efficacy of such models. Genomics-based knowledge and tools promise the ability to approach each patient as the biological individual he or she is, thereby radically changing our paradigms and improving efficacy.

In terms of prevention, for instance, testing that shows that an individual harbours a pathological mutation in a mismatch repair gene can lead to more aggressive surveillance and earlier surgical intervention to reduce the otherwise increased risk for colorectal cancer — individualized modifications in management that would make neither medical nor economic sense if they were applied to all patients. In terms of diagnosis, use of genetic microarrays is already making inroads in refining the...
• Biomedical researchers: Genome era; sequence of human genome; human haplotype map; etc etc

• For most clinicians the genome era has not yet arrived

• For genomics to have an effect on clinical practice: advances in the genomic literacy of health-care providers needed.
• Growing evidence that genomics will change the practice of medicine, but how????

• If there were an area of medicine that is appropriate for lifelong learning, it is genomics: young and quickly evolving

• We cannot .. discern.. clinical applications .. That will be commonplace in the working years fo today’s students and trainees.

• Teach knowledge, skills and attitudes that will make students lifelong learners of genetics and genomics.
What knowledge?

- Identify patients who require further investigation or specialist referral

- Understanding and communicating disease risk in order to facilitate and support the patient’s informed decision making.

- Modes of inheritance, the concept of multifactorial inheritance as well as awareness for rare Mendelian disorders.

- Low threshold for discussion with genetics specialist
What knowledge?

- Understand genetic tests, both (results of) molecular genetic tests to identify Mendelian disorders.... more common diseases .. such as breast cancer
- Understand information in lab report, including limitations
- Risk calculation <-> referral
- Pre-symptomatic testing for Mendelian disorders ... only in association with genetic counseling
- ..specific genes that contribute to specific disorders.. clinical significance.. distinction between abnormal genotype-fenotype.. implications
Genetische testen


Ook op de radio is voldoende aandacht besteed aan het onderwerp. In het radioprogramma De Ochtenden is op 30 november 2005 genuanceerd op dit onderwerp ingegaan.
Verilabs

Vaderschaps- en Verwantschapstesten

Verilabs

Met genetische tests geeft Verilabs helderheid inzake verwantschapvragen, DNA-profielen en (erfelijke) ziektebeelden. Verilabs' laboratorium beantwoordt vragen van consumenten en zakelijke en wetenschappelijke professionals op een eenvoudige, betrouwbare en betaalbare wijze.

Voor sommige tests wordt een thuistest toegestuurd en kunt u zelf monsters afnemen. Voor de meeste tests worden eerst intake gesprekken gehouden en wordt eventuele monstersname door ons op het lab uitgevoerd. Ook als arts, advocaat, notaris, overheid, (sport) manager of verzekeringsmaak kunt u de tests aanvragen en laten uitvoeren door ons lab of personen naar ons doorverwijzen.

Gewaarborgd door het ISO 17025-certificaat van zusterbedrijf BaseClear die de tests uitvoert, biedt Verilabs DNA-testen aan die voldoen aan de strenge eisen.
Essential skills

• Convey complex genetic information
  – Perception of risk
  – Attitude towards genetic testing
    • Cultural beliefs
    • Ethnicity
    • Personal experiences of disease in family
• Facilitate informed decision making
• Manage family dynamics
How will health-care professionals learn?

- Pre-service & in context
- Prevailing misconceptions (rare single-gene or chromosome disorders; genetic vs. non-genetic)
- .. convey to students and clinicians alike that genetics is qualitatively different from all other topics because it underlies all of pathophysiology, and is therefore the fundamental science of health and disease.
- .. argues implicitly against educational approaches that treat genetics solely as a medical specialty akin to all others.
What is needed?

• ..situate genetics in a practical clinical context

• ..inclusion of clinical examples to illustrate the fundamental principles of genetics as they relate to common diseases - .. occupy most of .. time..

• ..bridge gap between basic science courses and clinical perspectives...“think genetically”...
Recommendations

Box 4 | **Recommendations for integrating genetics into education**

- Integrate genetics across the pre-service curriculum
- Increase the amount of content that is related to genetics and common diseases, as opposed to rare Mendelian diseases
- Build bridges between basic sciences and clinical instruction
- Ensure that instruction is case-based and reflects practical examples that demonstrate that genetics matters on a daily basis and can improve patient outcomes
- Develop continuing-education programmes in conjunction with representatives of the target audience

Q7,8: as far as related to diagnosis, treatment, prevention of common diseases, which is more and more frequent...
• ..... combine new conceptual views of genetics with mechanisms that address those practical matters:

• Teach teachers

• Teach teams in which you collaborate (familial tumors, dysmorphology, clinics in other hospitals)

• Use internet

• Develop study material to be used more than once
Practical challenges

- Structure & sequence of curriculum

- Time constraints... busy clinical practice... only immediate needs of his or her patients..

- Any attempts to improve the quality and quantity of genetics that is taught to health-care professionals will need to combine new conceptual views of genetics with mechanisms that address those practical matters.
Deficiencies

• .. current educational approaches do not prepare students to practise in a health-care environment that will be increasingly influenced by genetics and genomics (*ref. 14-20; USA & EU*)

• Providers attitudes: .. Most providers view genetics as peripheral to everyday clinical concerns.

• The Human Genome Project is interesting, and I know that it will change health care dramatically some day.... unless.. tomorrow..I really don’t have time to deal with it.
Perceptions

- Genetics and genomics were not high on list of most important topics for continuing medical education GP’s... virtually every topic ...included relevant genetics content.
- Self-perceived lack of knowledge
- Misperception about reach of genetics
- Lack of self confidence in one’s knowledge
- Scepticism about the utility of current genetic applications.
Are we doing what we should do?

- Guideline adherence <100%
- Barriers:
  - Lack of awareness
  - Lack of familiarity
  - Lack of agreement
  - Lack of self-efficacy
  - Low expectancy of favourable outcomes
  - Inertia/lack of motivation
  - Perceived external barriers beyond control

Grol & Wensing. What drives change?.. MJA 2004;180:S57-60
Levels of Outcome of Educational Programmes
After Kirkpatrick, 1967 and Barr et al 2000

• Learners’ reaction
• Modification in attitudes and perceptions
• Acquisition of knowledge and skills
• Changes in behaviour
• Changes in organisational practice
• Benefits to users and carers