

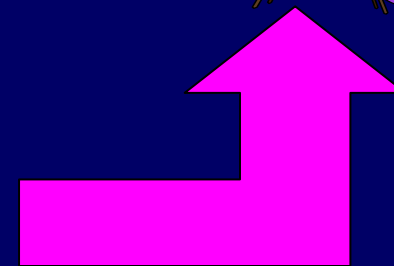
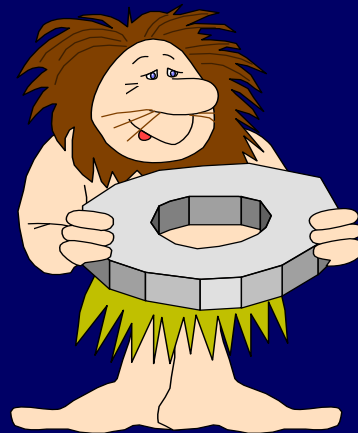
LABORATORY MEDICINE: ITS FUTURE

**Jocelyn M.B. Hicks, PhD, FRCPath
Professor Emeritus
The George Washington University
School of Medicine
Washington, DC, USA
President, IFCC**

DISCUSSION

◆ A few reflections on the
Past and Present

◆ Future

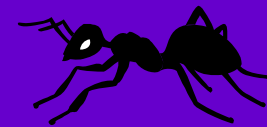


DISTANT PAST!

First tests known



Diabetes



Patient urinates on the floor. If the urine contains sugar, ants will crawl to lick the urine. This test was used up to 20 years ago in some parts of Africa



PHLEBOTOMY

- ◆ In the 20th century phlebotomy was introduced as a diagnostic tool
- ◆ Prior to that it was considered to be curative

EXAMPLES OF PHLEBOTOMY FOR CURING PATIENTS



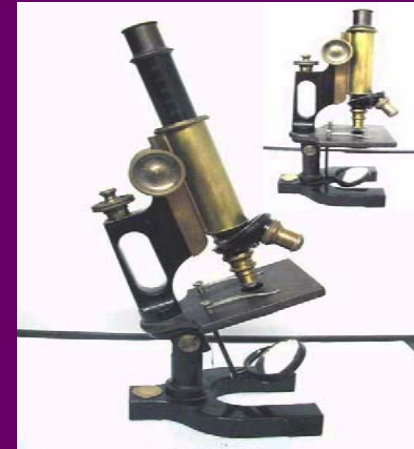
- ❖ 16th century Italy, physicians would order 15-20 leeches per hospital patient before examining them
- ❖ George Washington, the first US President, probably had only a common cold when he was bled daily as a treatment. It is thought that he died due to excessive blood loss



INSTRUMENTATION IN LABORATORY MEDICINE: 1920

A modern 200-300 bed hospital in the USA would be well equipped if it had. .

- ◆ A balance
- ◆ A microscope
- ◆ A centrifuge
- ◆ A Bunsen burner
- ◆ A Duboscq colorimeter

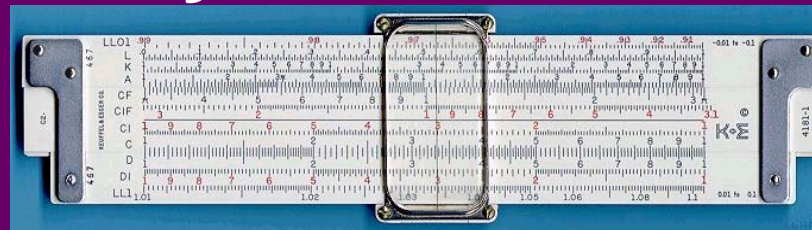


CLINICAL CHEMISTRY IN A HOSPITAL LABORATORY 1970

- ◆ Balance
- ◆ Spectrophotometer
- ◆ Flame photometer
- ◆ Van Slyke apparatus
- ◆ Klett colorimeter
- ◆ Centrifuge

IN 1970

- ◆ There were no calculators. Slide rules were used
- ◆ No automation
- ◆ No sophisticated quality control
- ◆ No fax machines
- ◆ No laboratory information systems



PRESENT

- ◆ **Point-of-Care Testing**
- ◆ **Molecular diagnostics**
- ◆ **Sophisticated equipment such as Tandem Mass Spectrometry**
- ◆ **Consolidation of testing on a single platform**
- ◆ **Consolidation of reference laboratories**



THE NEAR FUTURE

- ❖ **Short staffing**
- ❖ **Dramatic increase in POCT and home testing**
- ❖ **Non invasive testing**
- ❖ **Increased use of Tandem Mass Spectrometry**
- ❖ **Use of Molecular Diagnostics (Chips and SNPs), single cell analyses**
- ❖ **Use of robotics**
- ❖ **Working from home: telecommuting**

STAFFING PROBLEMS



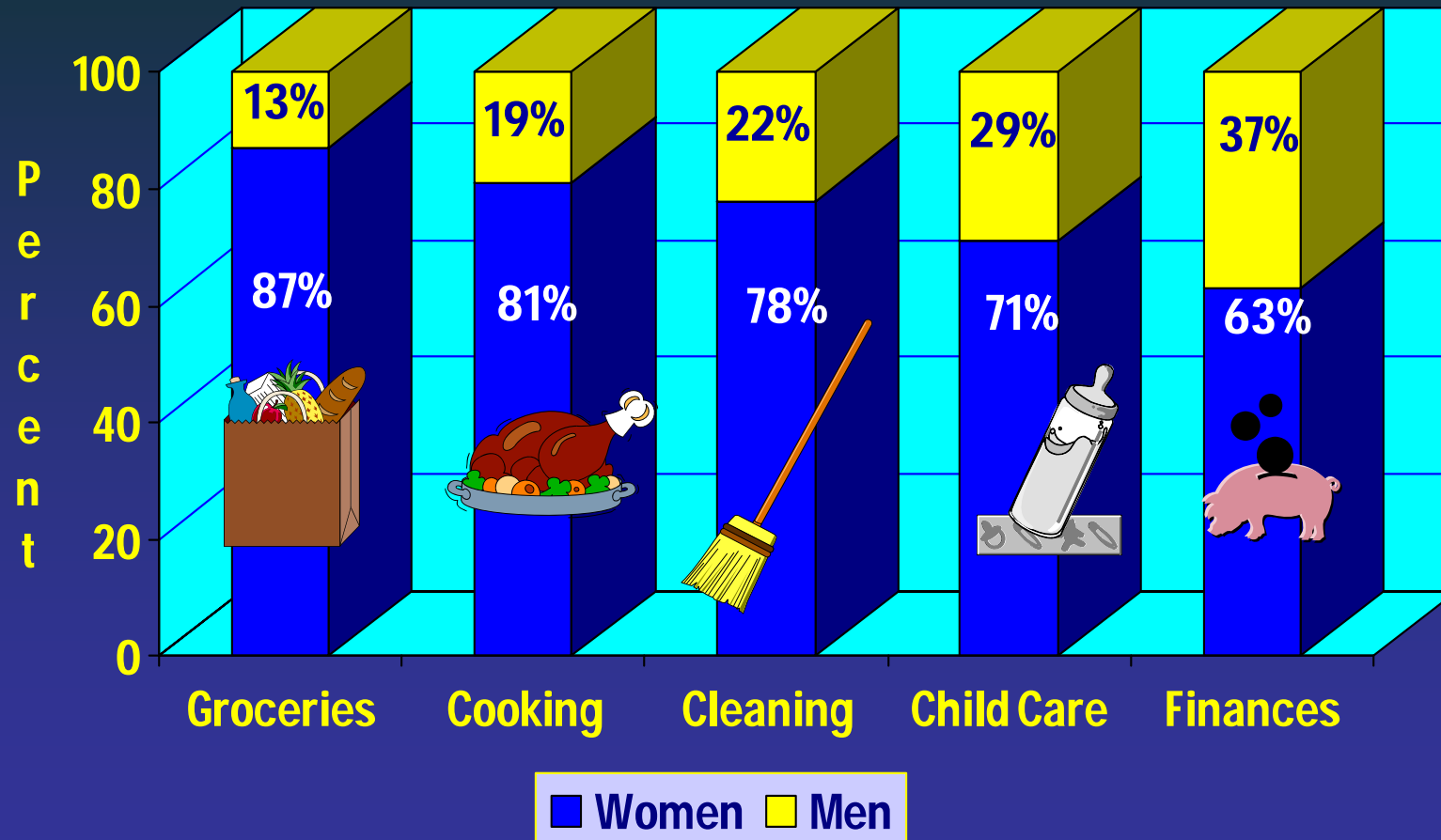
STAFFING PROBLEMS

**Staffing shortage of 13% nationwide in
the USA! *WHY?***

- **Medical technology schools closing**
- **Laboratory Medicine technologists
and technicians mostly women**
- **Women going into different fields**
- **Aging staff..average age nationwide
is 49y old**

DUAL INCOME FAMILIES (USA)

Home responsibilities



NON INVASIVE TESTING





GLUCOWATCH

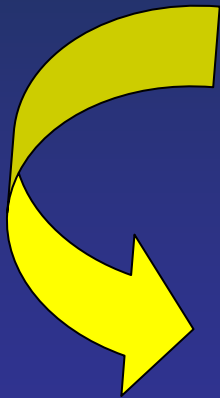
MOLECULAR DIAGNOSTICS



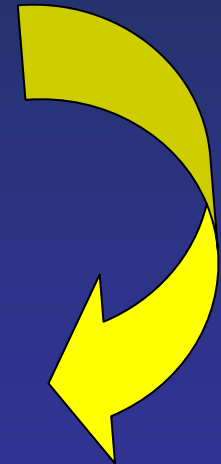
THE IMPORTANCE OF MOLECULAR DIAGNOSTICS

Lab results determine how 70% of healthcare dollars are spent

Molecular diagnostics is the fastest growing field within laboratory testing



Molecular diagnostics gives clinical practitioners more knowledge, better odds to fight and prevent disease



MOLECULAR DIAGNOSTICS: BETTER PATIENT CARE

- ❖ Infectious Disease & Resistance Testing
- ❖ Disease Prevention
- ❖ Personalized Medicine
- ❖ Technology Requirements:
 1. Certainty
 2. Control
 3. Consolidation

MOLECULAR DIAGNOSTICS

Single Cell Analyses

PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

**Offers an alternative to
traditional methods of
prenatal diagnosis including
chorionic villus sampling and
amniocentesis**

PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

**Allows genetic analysis and
selection of embryos to be
performed
prior to implantation and
pregnancy, and thereby
increasing the possibility of a
child free of Genetic Disease**

REQUIRES THE FOLLOWING STEPS...

- ❖ Production of embryos following a routine IVF cycle
- ❖ Growth of the embryos to ~8 cells (day 3)
- ❖ Biopsy (removal) of embryonic cells (blastomeres) for testing
- ❖ Capture of DNA (for PCR based tests) or intact nucleus (for FISH based tests)

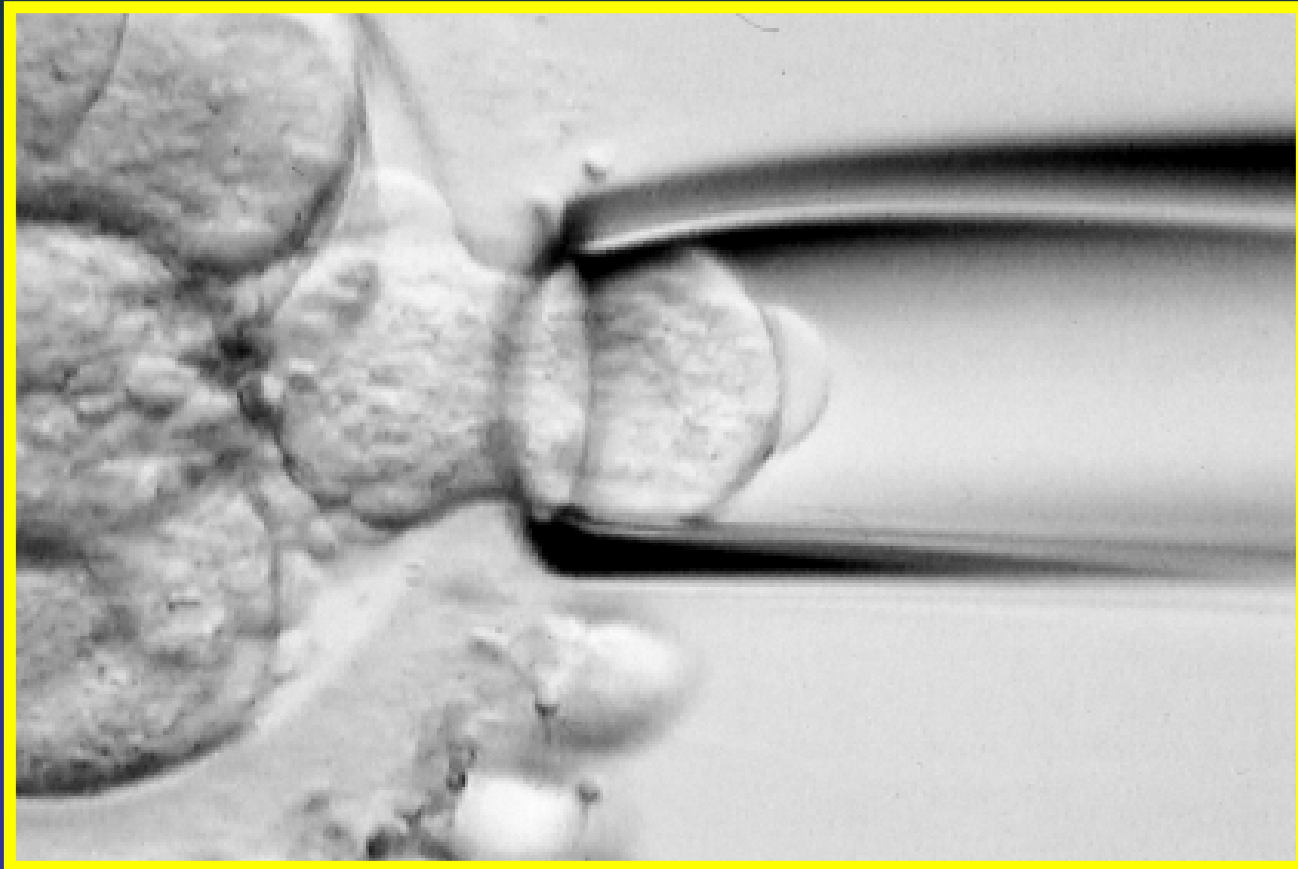
PGD:REQUIRES THE FOLLOWING STEPS...

- ❖ **Amplification of DNA (PCR based tests) or hybridization of fluorescently labeled DNA probes (FISH based tests)**
- ❖ **Interpretation and reporting of results**
- ❖ **Transfer of selected embryos into uterus on day 5 post retrieval**

EMBRYO BIOPSY



EMBRYO BIOPSY



PGD: NEW YORK TIMES SEPT 3, 2006

Article about a couple culling embryos obtained by in vitro fertilization to halt the strong heritage of Colon Cancer

- ❖ Is this unnatural selection or a wise decision in being sure their offspring does not carry the colon cancer gene?
- ❖ Is it a “pact with the devil” and “discriminatory” and producing designer babies, or is it right for persons to try to avoid deadly diseases in their progeny?

MOLECULAR DIAGNOSTICS

The Use Of DNA Chips

BENEFITS OF CHIP PLATFORMS

- **Combine all testing needs on one platform**
- **Cost-per-test decreases**
- **Test flexibility means lab can meet increasing test demands**
- **Technologist time is reduced**
- **One workstation means less bench space is occupied**

BENEFITS OF AUTOMATED CHIP TECHNOLOGY

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THE “-OMICS” REVOLUTION

✓ **Proteomics**

✓ **Pharmacogenomics**

✓ **Physiogenomics**

✓ **Nutrigenomics**

PROTEOMICS

- ❖ It is the large scale study of proteins, particularly their structure and functions
- ❖ The proteome is complex. It varies from cell to cell, and is constantly changing through its biochemical interactions with the genome and the environment
- ❖ The study of proteomics can lead to a better understanding of the disease process
- ❖ To catalog all human proteins is a major challenge for scientists. There is an international collaboration to achieve this goal that is being coordinated by the Human Proteome Organization

KEY TECHNOLOGIES used in PROTEOMICS

- One and two dimensional electrophoresis
- X-ray crystallography and magnetic resonance
- Tandem mass spectrometry
- Mass spectrometry
- Affinity chromatography
- X-ray tomography
- Software based image analysis

PHARMACOGENOMICS: THE LATEST!

- Pharmacogenetic tests can predict whether a drug will be effective or cause adverse, or even deadly side effects
- This especially applies to psychiatric and cardiac drugs
- Approximately 70 drugs have been identified that are catabolized by cytochrome P450 enzymes. There is now a test for these enzymes. More than 50 variations are known of the 2D6 gene that controls these enzymes

Pharmacogenomics, cont'd

- ❖ If there is too much of the enzyme, the drug will be catabolized rapidly, and will be less active. If the enzyme is reduced or absent, the drug will accumulate and produce the effects of an overdose
- ❖ 30% of persons of North African origin, 20% of persons of Middle East origin and 2% of Caucasians are born with 3 or more copies of the 2D6 gene causing extra rapid catabolism of certain drugs



PHYSIOGENOMICS: Hot off the Press!

Serum albumin-bound fragments: An archive of Potential Disease Markers

1. A protein fragment has been identified, which is derived from a protein encoded by the BRCA2 cancer associated gene
2. Protein markers have been identified for Alzheimer's Disease

1) Lowenthal MS, et al. Clin Chem 2005; 51:1933-45

2) Lopez MF, et al. Clin Chem 2005; 10:1946-54

NUTRIGENOMICS

- It is the field that examines the response of individuals to compounds in food using genomic and other related technologies
- “Nutrigenomics research looks at how diet interacts with gene expression”

NUTRIGENOMICS Cont'd

- ✓ **Identifying poor folate metabolizers**
- ✓ **Testing involves folate metabolism and the gene for 5,10-methylenedihydrofolate reductase (MTHFR). This enzyme converts 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate**
- ✓ **Mutations of the MTHFR gene are associated with homocystinemia, a risk factor for spina bifida births in pregnant women and premature cardiac disease**

NUTRIGENOMICS, Cont'd

- ✓ The ultimate goal would be to have broad-based population testing for health maintenance
- ✓ However before any testing becomes widespread it will have to be more evidence based
- ✓ A concern is could information gleaned from SNPS be misused by employers?

OTHER CHALLENGES FOR THE FUTURE

- **e-Technologies**
- **Global harmonization of IVDs**
- **Use of Nanotechnology**
- **Efforts to reduce Laboratory errors**
- **A effort to decrease unnecessary tests**
- **The changing population demographics in the US. Ethnic, cultural and racial diversity will change the incidences of major illnesses**
- **International competition in healthcare**



INFORMATION TECHNOLOGY(IT)



- **IT needs to be the backbone of healthcare**
- **It can lead to a better understanding of unnecessary tests**
- **It allows the development of evidence-based protocols**
- **Leads to an understanding of the “best” laboratory tests for the diagnosis of disease**

INFORMATION TECHNOLOGY

We are way behind!!

**20-25% of hospitals have computerized
physician order entry systems for
laboratory tests, or electronic medical
records**

WE MUST MOVE FASTER!



THE MORE DISTANT FUTURE, BUT COMING!!

📄 PERSONALIZED MEDICINE

📄 PREVENTIVE MEDICINE



NO LONGER "ONE SIZE FITS ALL!"

**PREVENTION:
*BETTER MEDICINE, BETTER
ECONOMIC SENSE***



- **Focus on early health rather than late disease**
- **It is better medicine to prevent disease early. e.g. treat cardiac disease at the onset of symptoms of high cholesterol, high blood pressure, etc.**

FACTS re NON- PREVENTIVE MEDICINE

- **Currently 70-80% of healthcare resources are spent on advanced diseases**
- **70 million baby boomers (age 50y and older) are eligible for colon cancer screening. Fewer than half have complied**
- **The 5 year survival rate for colon cancer is 90% for localized cancer and 8% if the cancer has spread further in the body**
- **Breast cancer survival has improved dramatically as a result of routine mammograms**

**Thank you CDC for
inviting me here today**

