

# The Future of Genomics: A Pediatric Perspective

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# Overview

- Promise of Genetic Technologies
- Application Paradigms
- Financial and Other Barriers
- Educational Challenges



# Looking into the Future



- Human Genome Project
- Genes mapped – not characterized
- Prevention through surveillance, life style changes or therapeutics
- Early identification provides the greatest opportunities as well as the greatest ethical challenges

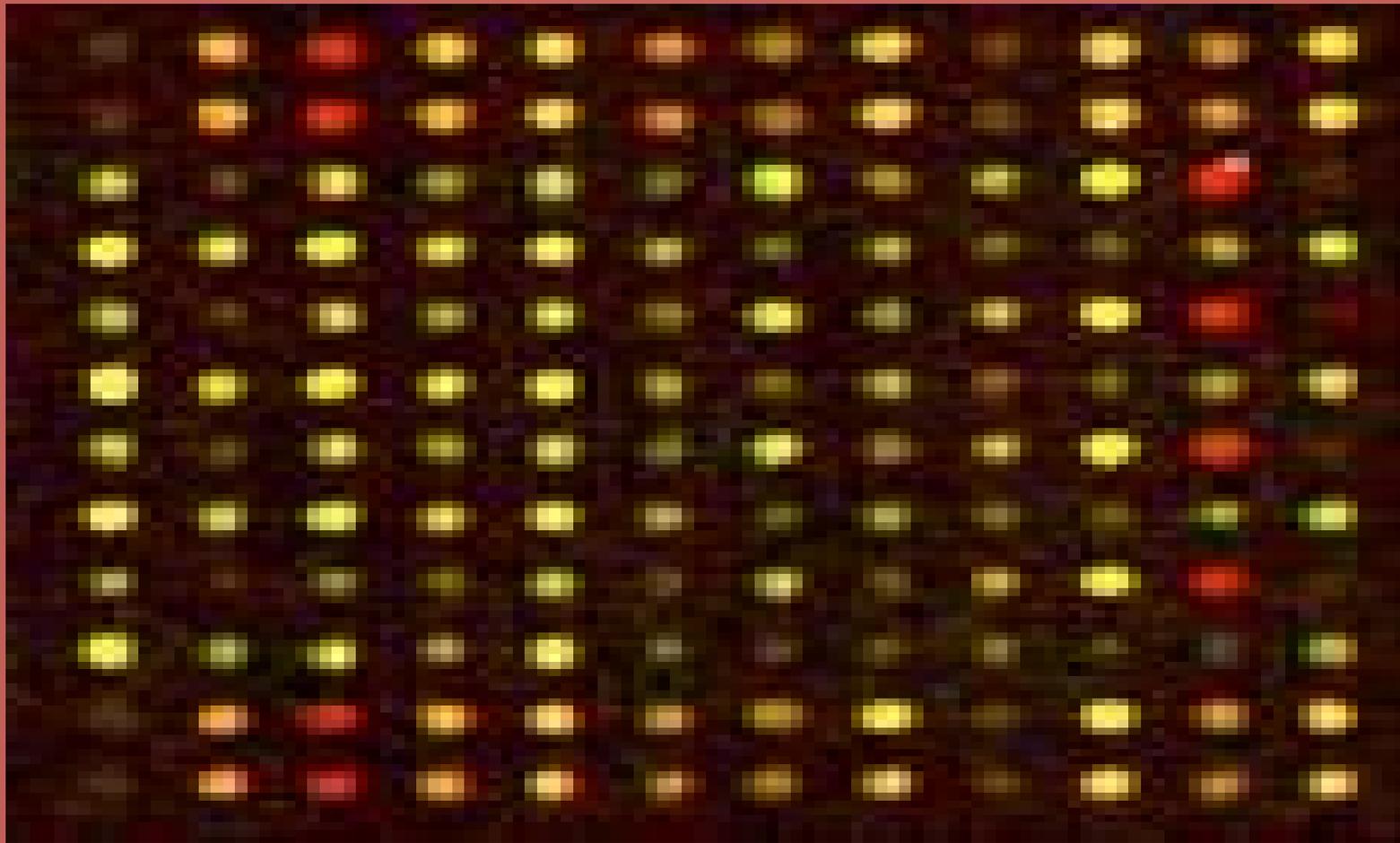


# Application of Genetic Technologies

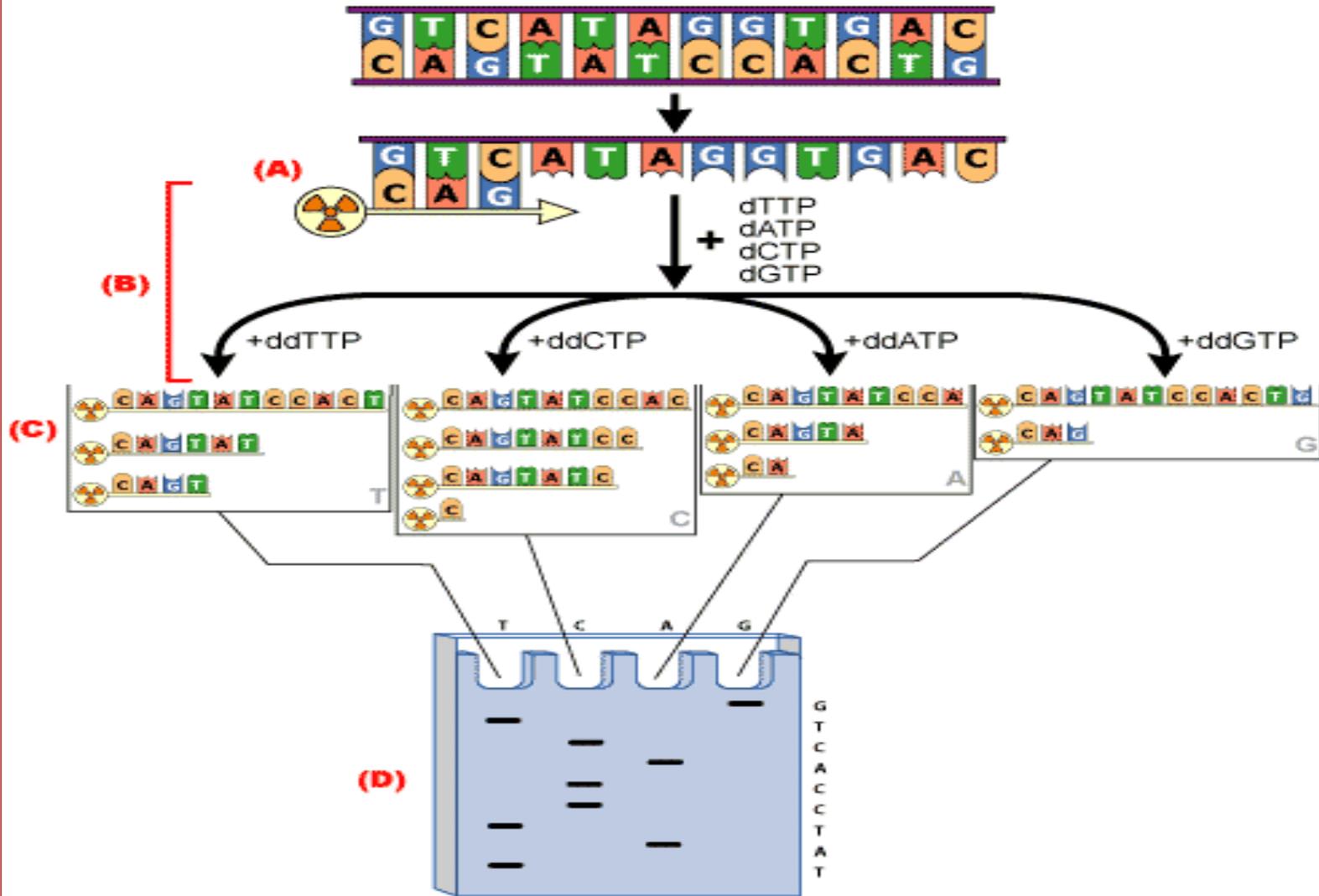
- Current practice – single gene disorders in patients with identifiable features
  - Limited scope and variably available
- Future practice – newborn screening model
  - General population screening for relevant conditions
  - Modifiable or treatable
- Future practice – pharmacogenetics
  - Use of genetic markers to provide guidance for medical management
  - Prevention of adverse events, selection of optimal therapeutics and individualized dosages



# CGH or Microarray Technology



# Next Generation Sequencing



# Barriers to Population Screening

- Limited knowledge about many genes
- Limited knowledge about gene-gene interactions
- Selection of conditions for which treatment or intervention is feasible
- Cost and infrastructure
- Workforce to interpret results and implement treatment
- Ethical considerations



# Workforce Challenges

- Pediatricians serving as head of the “medical home”
- Those who graduated from medical school before 1990 have limited exposure to new genetic technologies
- There were only 1,253 board certified clinical geneticists in the US as of 2007; that same year over 4.3 million children were born in the US
- That comes to 3,445 newborns per MD clinical geneticist



# How to Create a Useful Model?

- Pediatricians must remain as the center of the medical home
- Pediatricians need education about specific conditions on the gene chip
- Pediatricians need a genetic “hotline”
- Follow-up services should be in place before screening is begun
- Genetic experts to oversee the screening process and help with interpretation of screening results



# Basic Needs (Wish List)

- Panel of experts with a range of interests and backgrounds
- Cost effective, high throughput technologies for population screening
- Follow-up infrastructure and resources for primary care providers (PCPs)
- Well designed electronic medical records that are transportable
- Protected database for outcomes analysis
- Educational initiatives for PCPs
- Educational materials for parents



# Outcomes to be Measured

- Implementation of a screening process is of little use without ongoing assessment of clinical benefit
- Long term outcomes must be evaluated and may take many years to demonstrate benefit or no benefit
- Thoughtful introduction of new genes on the chip requires broad expertise and general consensus



# Cost Conundrum

- Current reimbursement system is not set up to handle single gene sequencing, let alone general population screening
- Newborn screening costs fall primarily to hospitals who “purchase” filter papers for testing through their state
- Indirectly, these costs are passed on to insurance companies and/or states which collect premiums and taxes
- Ultimately the cost is born by employees and taxpayers



# Moral Obligation

- Do whatever is necessary to ensure that:
  - Tests are appropriate and are likely to promote health benefits
  - Results cannot or will not be used in a way that will disadvantage a gene carrier in terms of educational opportunities, insurance coverage, medical care, or employment
  - Ongoing assessments are made to add tests that show promise for improved health and delete tests that are shown to be of little value
  - The financial burden of testing is mitigated by health benefits for all children born in the US



# Closing Thoughts

- Money spent on prevention and good health for children has the potential to reap benefits far in excess of any other investment we make as a nation
- Whereas the future of our country may depend on a healthy adult workforce, we need to do everything in our power to ensure that genetic technologies are used wisely and equitably



