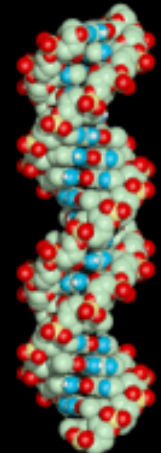
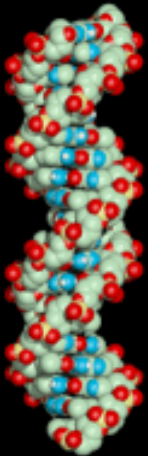


The Promise of Personalized Medicine

Where Are We?








Paul G. Rothberg, Ph.D., FACMG

Director, Section of Molecular Diagnosis

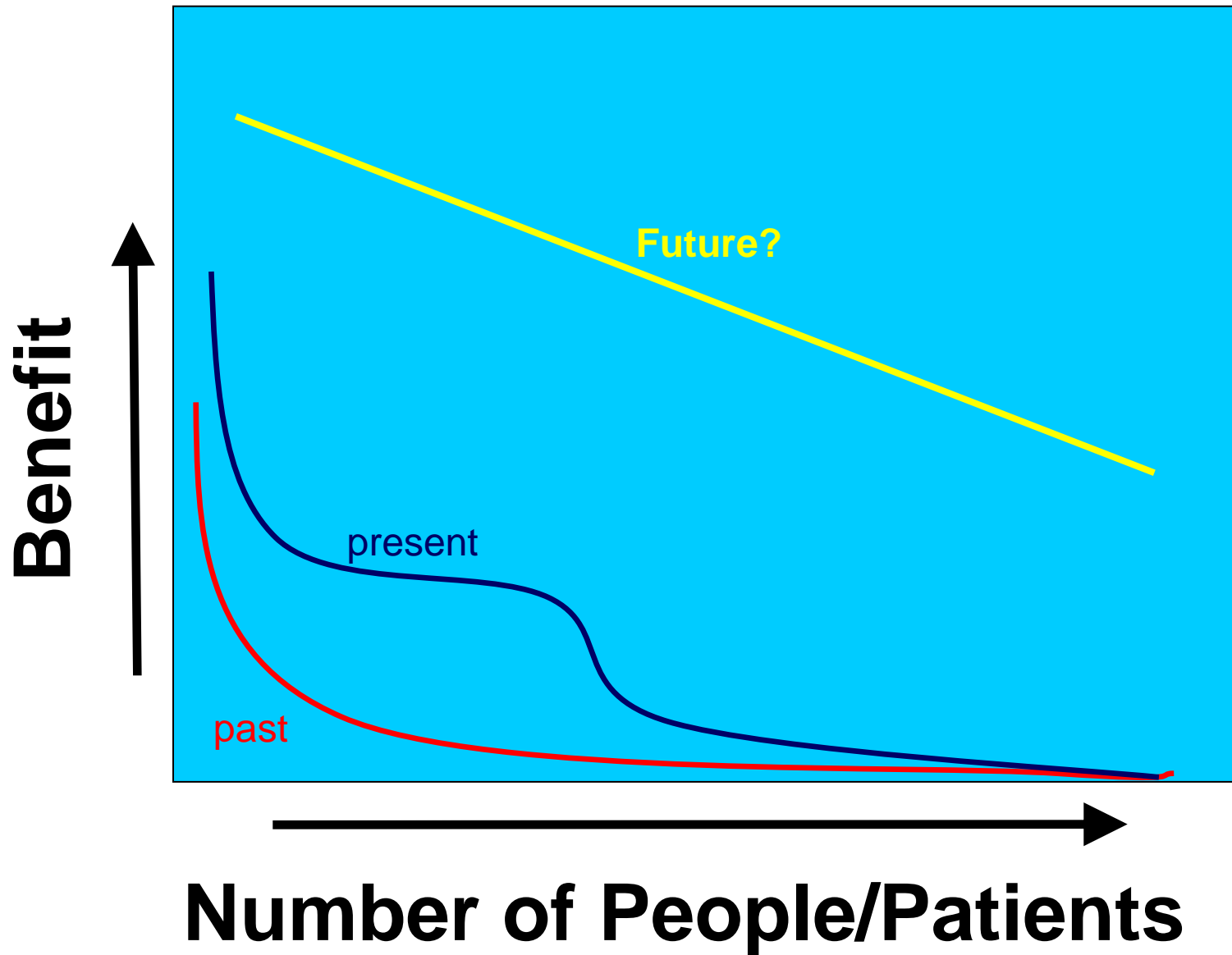
URMC Labs

What is Molecular Diagnosis?

-  **Inherited Disease (molecular genetics)**
germline
-  **Molecular Microbiology**
biggest part of MD
-  **Molecular Oncology**
somatic
-  **Pharmacogenetics & Personalized Med.**
back to the germline
-  **Direct to consumer**
marketing > science



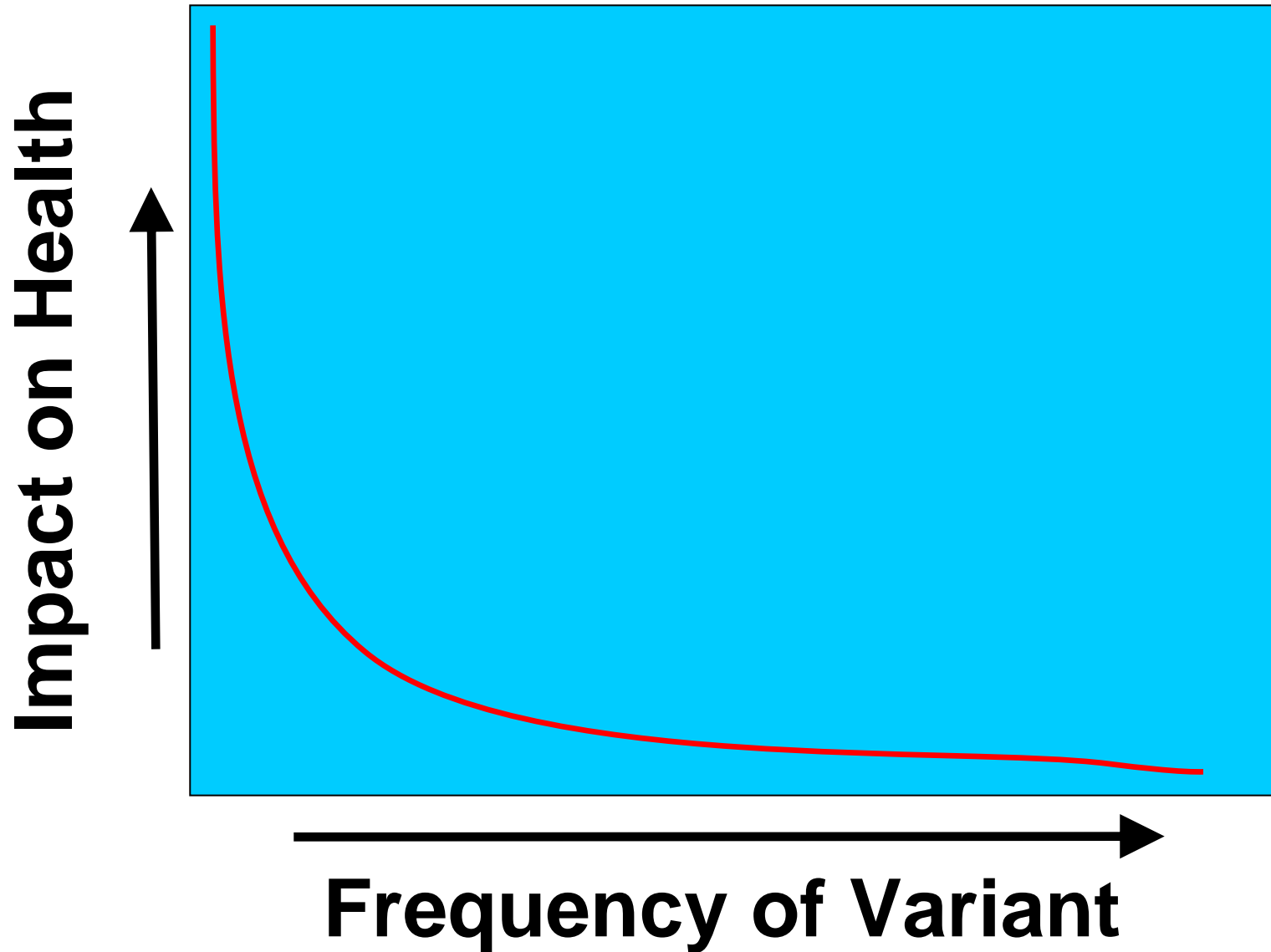
Benefits from Molecular Diagnosis






"Here's my
sequence..."

Health Impact of Genetic Variation







How to Make PM Real

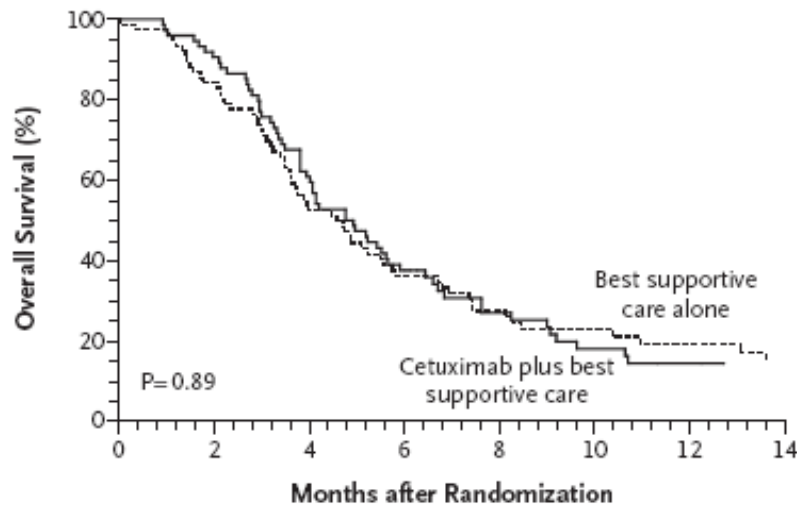
- 🧬 Bring down the cost of analysis
- 🧬 Increase knowledge of health effects of DNA variation (epistasis)
- 🧬 Find Ways to Use this Information;
Clinical Utility 
- 🧬 Delivery of PM; Education

New Test: **Detection of KRAS Mutation in Colorectal Cancer**

Features of KRAS Mutation Testing

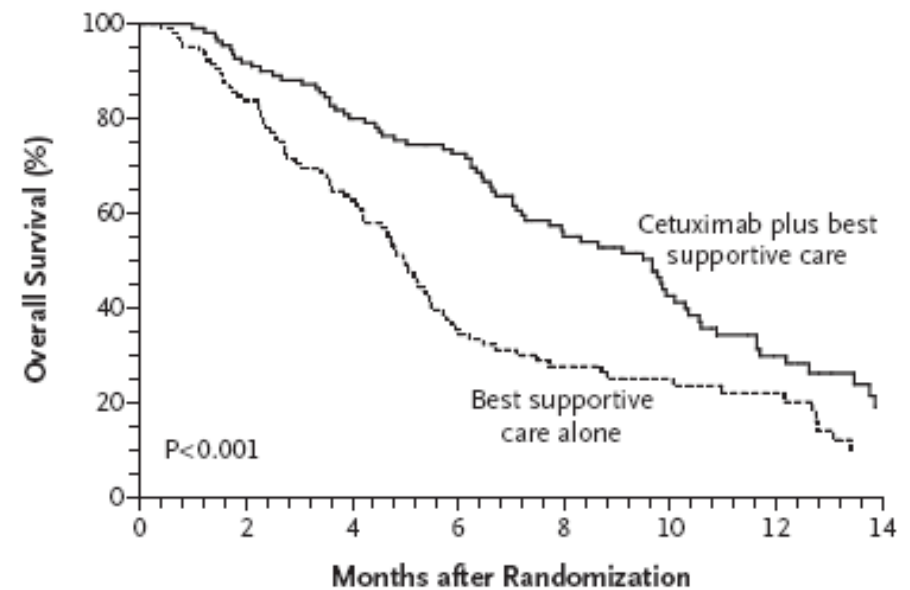
-  Mutation is present in 30-50% of colorectal CA (1980s).
-  Predicts lack of response to anti-EGFR monoclonal antibody therapy (recent discovery).
-  Saves expense & toxicity of ineffective therapy.
-  Not all tumors with normal KRAS respond to anti-EGFR therapy. Better predictive testing needed.

Overall Survival for Patients Comparing Cetuximab with BSC



WITH
← **KRAS Mutation**

WITHOUT
→ **KRAS Mutation**



Ref: Karapetis, et al. 2008,
NEJM vol. 359, p. 1757.