

# New Developments in Precision Medicine : Implications for Practice and Policy

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## What is Precision Medicine?

**“An emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle”**

(US National Institutes of Health )

- Ability to predict more accurately which treatment and prevention strategies will work for whom allows us to (i) improve outcomes and (ii) avoid adverse effects or treatment toxicities
- Similar to “personalized medicine”, but not focused on treatment regimes developed uniquely for each individual, rather on identifying which approaches will be effective for which identifiable groups of patients based on genetic, environmental, and lifestyle factors.



# Pharmacogenomics

Individual's susceptibility to certain diseases or response to a treatment are sometimes linked to specific common DNA variations, or single nucleotide polymorphisms (SNPs)

Pharmacogenomics can tell us about specific genes encoding either metabolic enzymes or defective structural proteins which lead to

- The need for a higher dose to achieve a therapeutic effect
- A greater risk of side effects or more severe side effects
- Variation in effect size or likely benefit from the treatment



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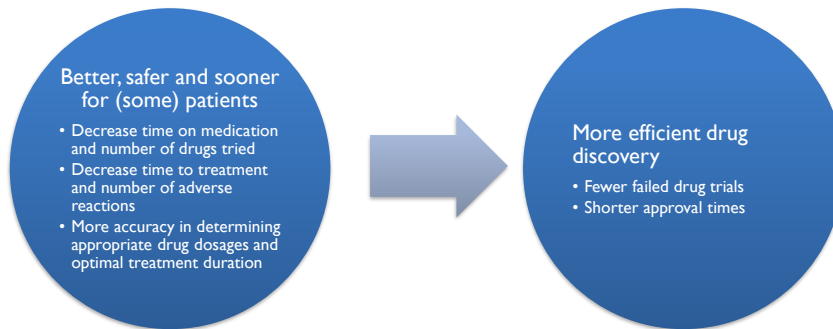
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**Example : Acute myeloid leukemia (AML)**

- Combinations of various doses and schedules of drugs used as treatment for all types of AMLs (of which there are many)
- Significant variation in responses, some of which are very severe adverse reactions
- Prognostic and predictive biomarkers can identify patients who could benefit from a particular treatment or those exhibiting higher risks of toxicity.

# Potential for Patients And Pharma



## Peril as Well...

- Field is still in early stages, unclear that most “high-tech” discoveries are sufficient to be clinically meaningful
- Need to look beyond genetics towards a systems approach, but adding biological, environmental, and behavioral data = massive conceptual and computational problem
- Cost of genetic sequencing is falling but still high
- Cost of drugs may stay high with potential for price-discrimination
- Opens up new ethical and practical considerations for practice

# Ivacaftor

**KALYDECO® (ivacaftor)**  
oral granules for children  
age 2 years to less than 6

[Learn more](#)

**What is KALYDECO?**  
KALYDECO is a prescription medicine used for the treatment of cystic fibrosis (CF) in patients age 2 years and older who have one of the following mutations in their CF gene: G551D, G1244E, G1349D, G1778R, G551S, S1251N, S1255P, S549N, or S549R. KALYDECO is used for the treatment of CF in patients age 2 years and older who have an R1177H mutation in their CF gene. KALYDECO is not for use in people with CF due to other mutations in the CF gene. KALYDECO is not effective in patients with CF with two copies of the F508del mutation (F508del/F508del) in the CF gene. It is not known if KALYDECO is safe and effective in children under 2 years of age. Please see Important Safety Information below.

Can reverse disease in patients with a specific mutation in the CF gene

Fast-tracked by the FDA

## Cost-effectiveness is not guaranteed

- Costs up to \$300,000 a year per patient
- For patients with suitable mutation, recent evidence suggests impact equal to that of three universal (and cheaper) treatments: high-dose ibuprofen, aerosolized saline and the antibiotic azithromycin
- At present, does not help 95% of the entire CF patient population

### Vertex (VRTX) To Stop VX-661-Ivacaftor Study, Stock Down

August 10, 2015, 10:20:00 AM EDT by Investor's Business Daily

[Comment](#)



Vertex Pharmaceuticals Incorporated's VTX shares were down in pre-market trading on news that the company will be stopping one of the phase II studies being conducted on a VX-661-ivacaftor (trade name, Kalydeco) combination.

The company announced that it will not be continuing the phase II study in patients with one copy of the F508del mutation and a second mutation that results in minimal CFTR function. The decision was based on a planned interim-futility analysis of the first part (part A) of the two-part study. The analysis by an independent Data Safety Monitoring Board (DSMB) showed that VX-661 plus ivacaftor did not result in a pre-specified improvement

## What does the future hold?



Is our current  
commitment to research  
+ regulation enough to  
ensure a  
“transformational leap” ?