US Food and Drug Administration

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Basics

Disclaimer

- The views in this presentation are my own
- I have no commercial involvement in any of these activities

What I will cover today

- FDA and Precision medicine
- Federal initiative
- What we have done so far

Traditional approach: One size

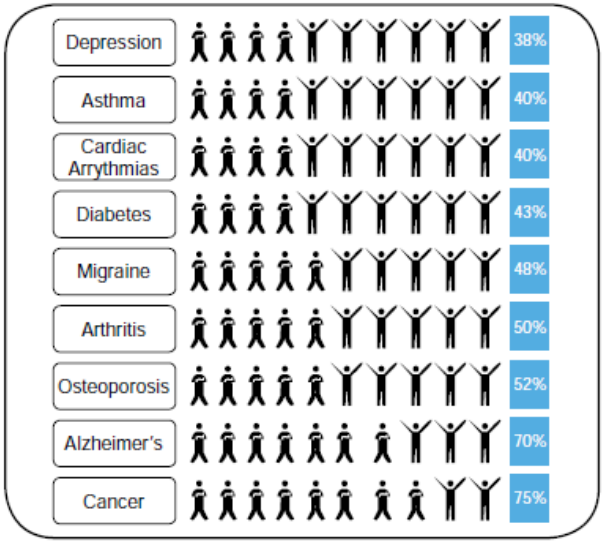


Figure 2. Percentage of patients for whom drugs are ineffective. (Source of data: Spear, B.B., Heath-Chiozzi, M., & Huff, J. (2001). Clinical application of pharmacogenetics. *TRENDS in Molecular Medicine, 7*(5), 201-204.) (Note that lack of efficacy in a given patient may reflect a complex interaction of factors and can also result from inadequate or inappropriate dosing regimens of a drug that would otherwise be effective, as well as lack of adequate patient compliance.)

Precision Medicine Initiative (PMI)

Program to empower patients, researchers, and providers to work together toward development of individualized treatments

PMI Objectives

- More, better treatments for cancer
  - Identify more genomic drivers to help develop more effective treatments
- Develop 1M person voluntary national research cohort
  - A patient-powered research effort that leverages existing research and clinical networks
  - Development of interoperability standards and privacy protections to enable data exchange and sharing
- Protect patient privacy

PMI Objectives (continued)

- Regulatory modernization
  - Streamline regulatory processes for Next Generation Sequencing (NGS) technologies
  - Enable patient access to their own health information and the software needed for its safe and accurate analysis
- Foster public-private partnerships
  - Develop infrastructure to expand cancer genomics knowledge
  - Launch the voluntary research cohort

Tailor Health Care to Each Patient

The right treatment

The right patient

The right time

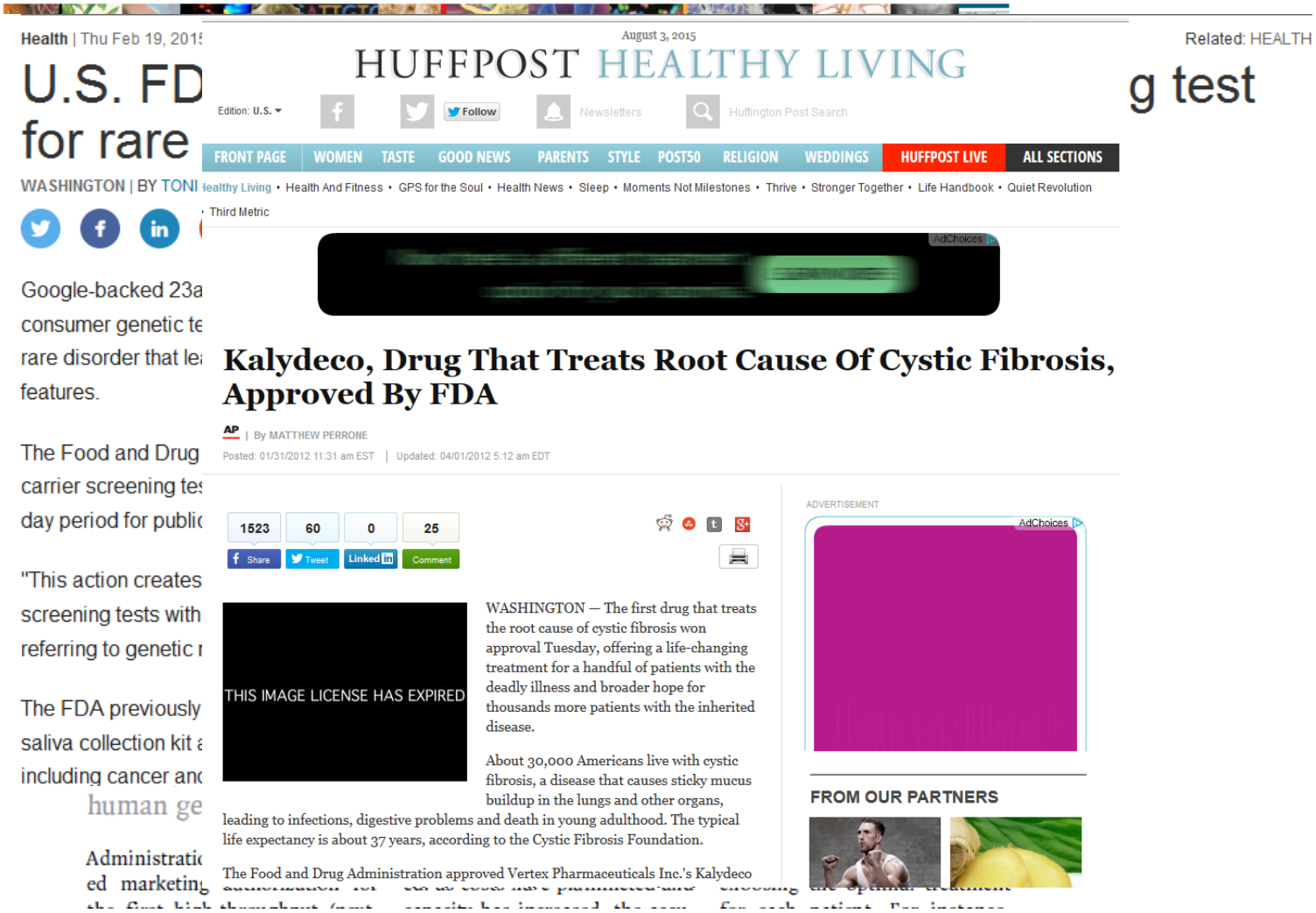
Timing is Everything: Build and recycle

- Advances in understanding of disease
  - Large-scale genome-based research cohorts established across the world
  - Human-genome sequencing continues to get cheaper and faster
  - Availability of new data – microbiome, diagnostics, and sensor data
- Clinical applications based on genomics and other biomarkers are now available and being used in the care of patients
- Every new bit of data adds to the picture, every new test method advances knowledge to build upon

What has been done?

- Over 23 companion diagnostics cleared or approved
- Over 50 biomarkers used in targeting 147 approved drugs
  - Cystic Fibrosis, Cancer, Cholesterol, Psychiatric, Pulmonary, Infectious Diseases, etc.
- Over 56 approved/cleared human molecular tests
- More than 24 relevant Guidances issued in past decade

Examples of building blocks



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August 3, 2015

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Kalydeco, Drug That Treats Root Cause Of Cystic Fibrosis, Approved By FDA

AP | By MATTHEW PERRONE

Posted: 01/31/2012 11:31 am EST | Updated: 04/01/2012 5:12 am EDT

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WASHINGTON — The first drug that treats the root cause of cystic fibrosis won approval Tuesday, offering a life-changing treatment for a handful of patients with the deadly illness and broader hope for thousands more patients with the inherited disease.

About 30,000 Americans live with cystic fibrosis, a disease that causes sticky mucus buildup in the lungs and other organs, leading to infections, digestive problems and death in young adulthood. The typical life expectancy is about 37 years, according to the Cystic Fibrosis Foundation.

The Food and Drug Administration approved Vertex Pharmaceuticals Inc.'s Kalydeco

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FDA Guidances: Standards matter

- Standards for Next Generation Sequencing (NGS)-based in vitro diagnostic tests (IVD) used to diagnose certain hereditary diseases
  - How FDA will assess test devices before marketing
  - We need strong standards, but flexible approaches
- Using public genetic databases to support validity of NGS-based IVD
  - How to make sure data can support rapid marketing approval
  - How large databases can improve test interpretations and use
  - The better the information, the more applicable the tests for patient use

Guidance is not enough

Modernizing regulation: Next Generation Sequencing testing

- New regulatory strategies for NGS
  - Standards to ensure quality
  - ***Open-source tools*** to help test developers meet standards
  - Promote translation and innovation by adopting a ***dynamic regulatory system***

- How we are doing this
  - FDA Workshops
  - Partnerships
  - Calls for research proposals

FDA Sponsors Key Projects: Stanford CERSI*

- Scientific training course
  - Challenges of analyzing genome sequences
  - Introduce **precisionFDA** portal
  - exercises for researchers on portal use to understand best practices and help improve them
- Research on ancestry genomics in analysis of diagnostic test methods
- Analysis of sequencing technology - consistency and reproducibility

*Center for Regulatory Science and Innovation

PrecisionFDA Portal: How it works

- FDA sponsors portal for researchers to share
- Open platform
  - Any scientist or research organization can register
- Access to data, methods, a way to benchmark, share and improve
- Fostering innovation

precisionFDA

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precisionFDA

A community platform for NGS assay evaluation and regulatory science exploration.

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NOTES

- **Write and publish** rich notes describing your thoughts and your work
- **Attach** any files, comparisons, or apps
- **Read** what others are reporting and reproduce their workflows

Learn more

FILES

- **Upload** your own files on cloud storage
- **Generate** files through running apps
- **Publish** reference data or any other files
- **Browse** other members' contributions

Learn more

COMPARISONS

- **Quantify** the similarity between two sets of genomic variants (VCF files)
- **Compare** your own test set (for a given biospecimen) to established benchmark sets

Learn more

APPS

- **Run** mapping and variation calling pipelines or other Linux-based software apps on the cloud
- **Contribute** your own software and scripts and let others explore them

Learn more

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https://www.youtube.com/watch?v=ir662wQJ5cI

precisionFDA Challenges

App-a-thon in a Box

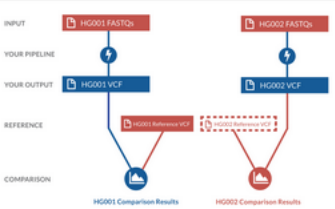
28 responses · 79 followers

- Challenge started 31 Aug 2016 12:00 UTC
- Submissions closed 31 Dec 2016 23:59 UTC
- Results announced 04 Jan 2017 22:10 UTC

Truth Challenge

25 responses · 59 followers

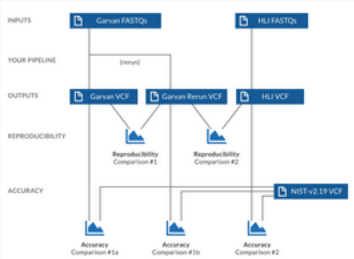
- Challenge started 27 Apr 2016 03:59 UTC
- Submissions closed 27 May 2016 03:59 UTC
- Results announced 29 Jun 2016 13:30 UTC



Consistency Challenge

17 responses · 54 followers

- Challenge started 26 Feb 2016 03:59 UTC
- Submissions closed 26 Apr 2016 03:59 UTC
- Results announced 26 May 2016 00:50 UTC



Latest News and Announcements

The App-a-thon in a Box Challenge is now closed!

January 3rd, 2017

We would like to thank all the

Thank you for sharing your experience!

December 19th, 2016

PrecisionFDA would like to thank its

precisionFDA MEMBER

Precision Medicine

- We are at the beginning – success is sweet
  - Individual cancer tissue genomics
  - Metabolism markers
- Next Generation Sequencing (NGS)
  - Use in diagnostics will launch great strides
  - Standards in assessment, tool development and validation are critical
- Regulators must be engaged at outset
  - Technology is fast and we must be nimble
- FDA is investing and invites anyone to join in