The pioneering role of rare diseases in science

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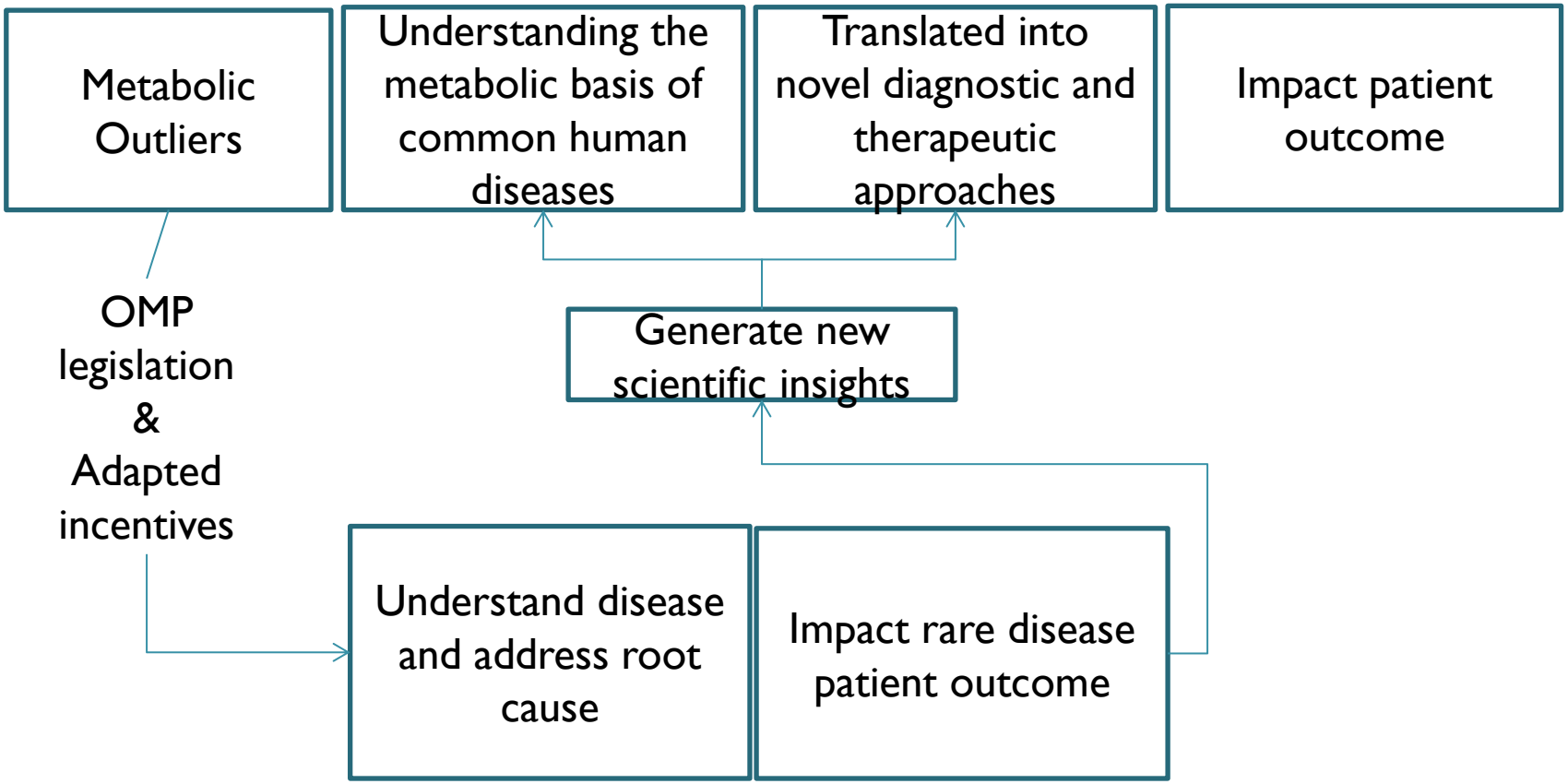
efpia
European Federation of Pharmaceutical Industries and Associations

EuropaBio
The European Association for Bioindustries

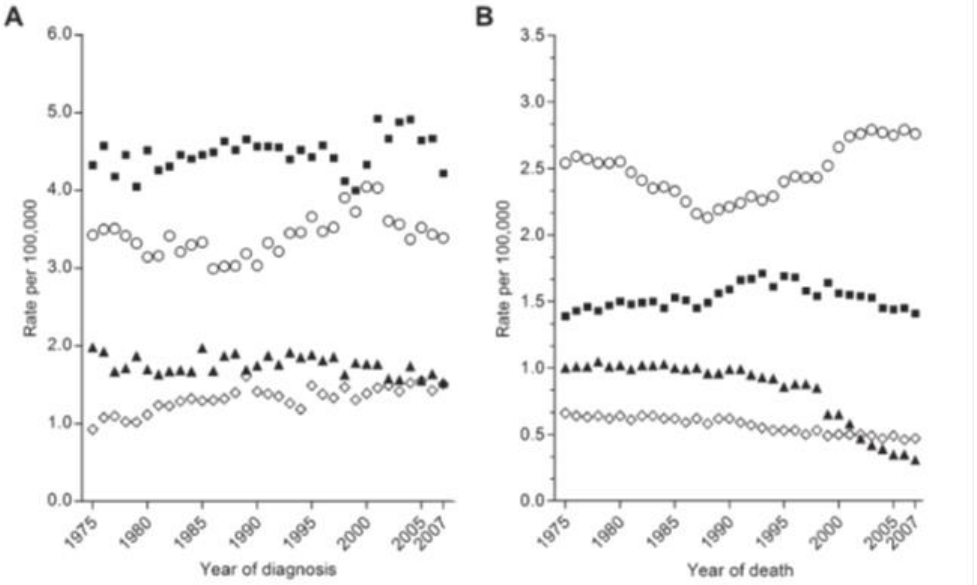
Treasure the exceptions
Open new paths



Chronic Myeloid Leukemia:

Step change treatment

Figure 2. Surveillance, Epidemiology, and End Results (SEER) incidence and mortality rates in chronic lymphocytic leukemia (**black squares**), acute myeloid leukemia (**white circles**), chronic myeloid leukemia (**black triangles**), acute lymphocytic leukemia (**white diamonds**) between 1975 and 2007. **A**) Age-adjusted incidence rates by cancer site for all ages, all races, and both sexes. **B**) Age-adjusted US mortality rates by cancer site for all ages, all races, and both sexes. Rates are per 100 000 and are age-adjusted to the 2000 US standard population. From the SEER database (SEER 9) (http://seer.cancer.gov/faststats/selections.php?run=runit&output=1&data=1&statistic=1&year=201001&race=1&sex=1&age=1&series=cancer&cancer=92;93;96;97 and http://seer.cancer.gov/faststats/selections.php?run=runit&output=1&data=2&statistic=1&year=201001&race=1&sex=1&age=1&series=cancer&cancer=92;93;96;97).



A decade ago, the treatment of CML had been dramatically changed by tyrosine kinase inhibitors (TKIs)

Before the introduction of this novel medicine, only 30 percent of CML patients survived to 5 years after diagnosis. Nowadays the overall survival rate at 5 years is 85%–95%.

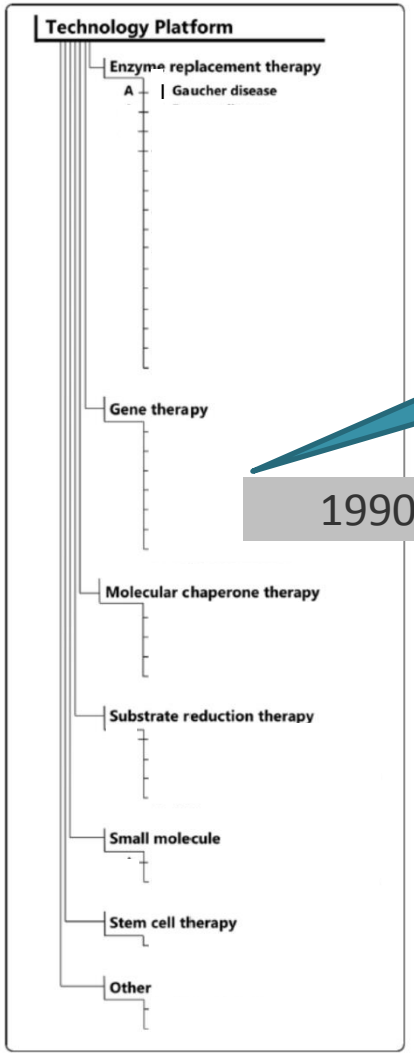
Gambacorti-Passerini C et al. Multicenter independent assessment of outcomes in chronic myeloid leukemia patients treated with imatinib, J Natl Cancer Inst 2011;103:553-561

LEUKEMIA NET : http://www.leukemia-net.org/content/home/index_eng.html [consulted May 2015]

Baccarani M et al. Chronic myeloid leukemia: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up, Annals of Oncology 2012; Supp 7:vii72–vii77

Lysosomal storage disorders: 40 diseases

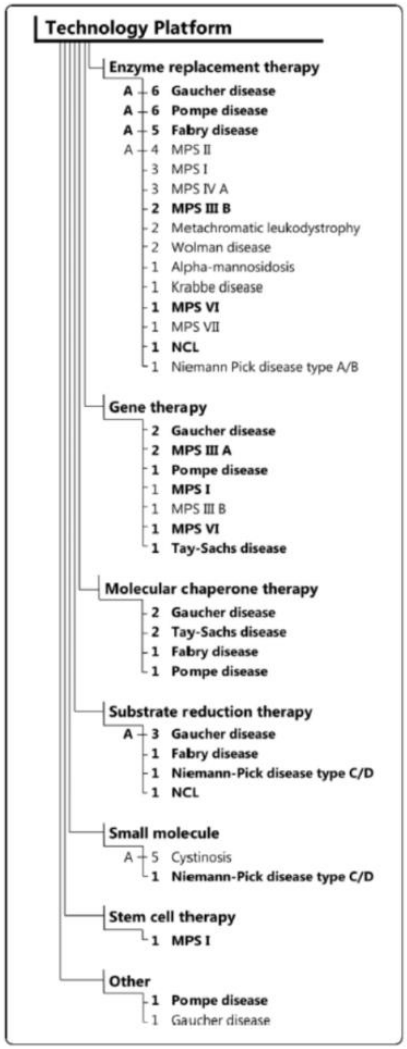
Enzyme replacement therapy for fatal genetic diseases



Alglucerase
First placental-derived glucocerebrosidase ERT approved

1990 2000 2010 2012 2013

14 approved treatment for 7 diseases
7 different technological platforms



Mechler K , Mountford W, Hoffman G, Ries M. Pressure for drug development in lysosomal storage disorders – a quantitative analysis thirty years beyond the orphan drug act. Orphanet (2015) 10:46

Patient early diagnosis and survival

Unmask disease predispositions and links with other disease mechanisms

- Cancer and Inborn Errors of Metabolism

Hypertyrosinemia type I

"The use of nitisinone in hypertyrosenemia type 1 significantly reduce the need for liver transplantation and the risk of developing liver cancer in patient surviving after 2 years of age"

- Neurodegenerative and Genetic diseases

Gaucher disease

Since treatment for Gaucher patients is available for 25 years. Gaucher is now a well-studied disease and patients and carriers appear to be at higher risk for P.D. *"A better understanding of the link between Gaucher and Parkinson Disease may shed light on what causes Parkinson Disease in general.."*

Erez, A., Shchelochkov, O.A., Plon, S.E., Scaglia, F., and Lee, B. (2011). Insights into the pathogenesis and treatment of cancer from inborn errors of metabolism. Am. J. Hum. Genet. 88, 402–421.

Parkinson disease fondation : http://www.pdf.org/en/science_news/release/pr_1400266646 [consulted May 2015]

The speed of science is often excruciatingly slow

OPINION

The most transformative drugs of the past 25 years: a survey of physicians

Aaron S. Kesselheim and Jerry Avorn

26 drugs or drug classes were selected
- 10 had an orphan designation by the FDA. Most of the designation by the FDA dated before the OMP legislation was enacted in Europe .
- 2 of the 10 had also a designation in Europe.

Kesselheim A, Avorn J. The most transformative drugs of the past 25 years: a survey of physicians Nature Reviews Drug Discovery 12; 425-431 (2013)

Kesselheim A , Tan Y , Avorn J. The roles of academia, rare diseases and repurposing in the development of the most transformative drugs. Health Affairs 34 ; 286-293 (2015)

A look to the future

New technology platforms

- Gene therapy
- Regenerative medicine

Methods and processes

- At what point does careful clinical observation cease to be science?

Expectations and Time

- There is genuinely great rare disease research that produced improved patient outcome in serious diseases
- We are not done yet and have great expectations for the next 15 years.
- The speed of science is often excruciatingly slow
- There is a significant lag time to be able to fully grasp the impact on health outcome and clinical practice