

Rare Disease Clinical Research Networks in the USA

Working Together Makes a Difference

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My Own Experience

Urea Cycle Disorders: A Tale of Three Patients and Cooperation



1980s

< 5%



1990s

< 50%

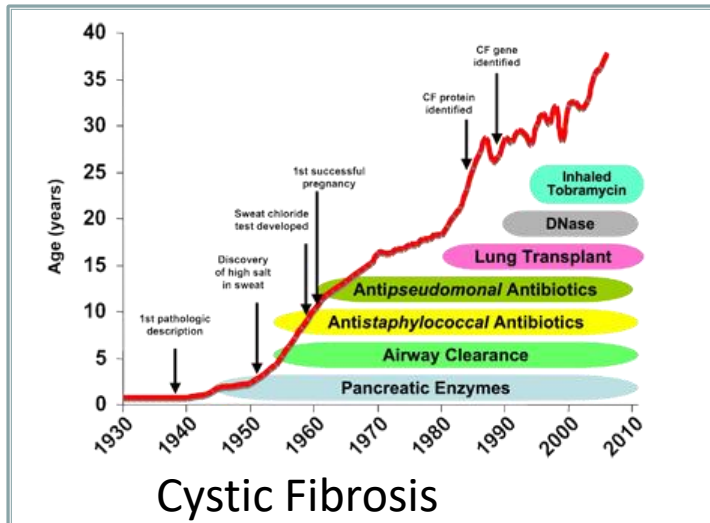
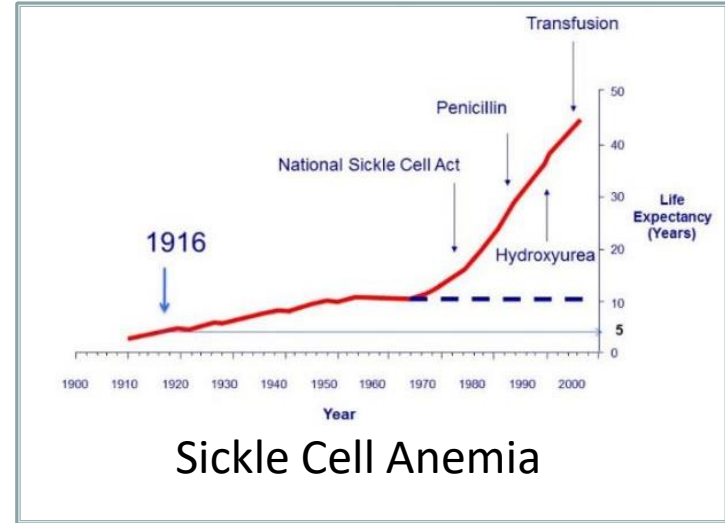
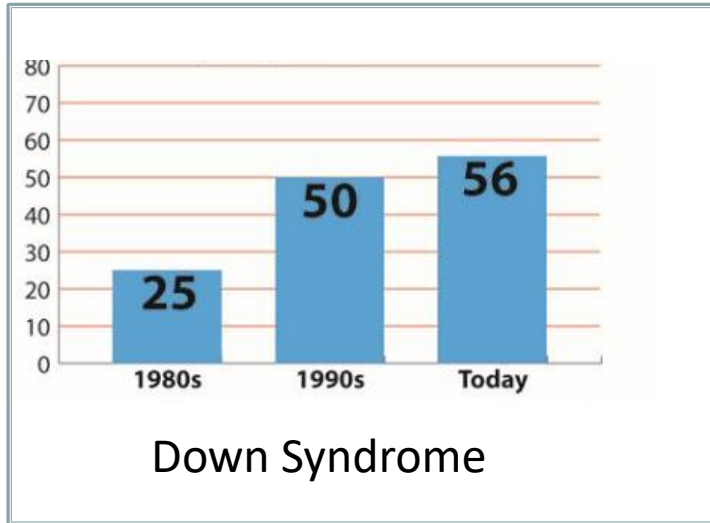


2017

> 95%

Without a substantial new therapy!

It is a trend



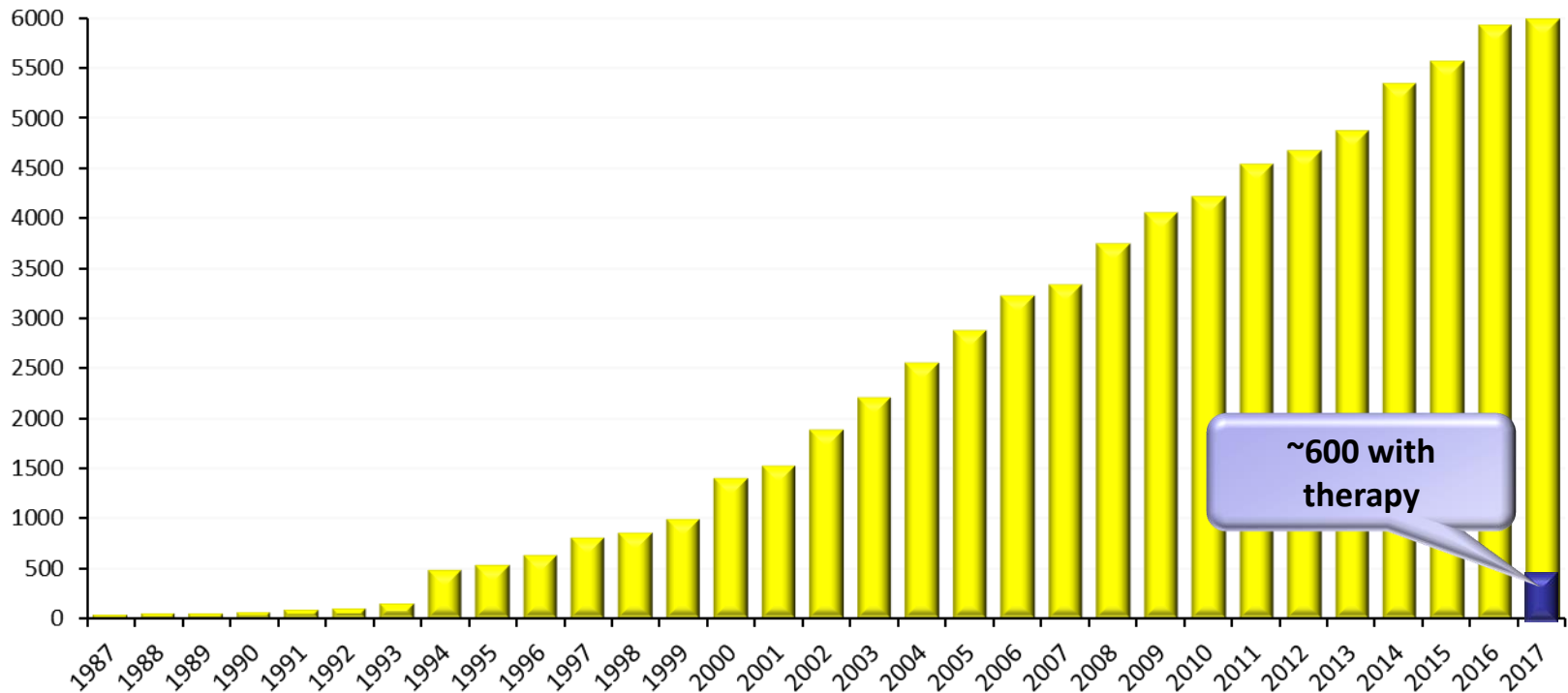
All of these effects are primarily the result of collaborating on standards of care and new therapeutic development through sharing information.

Rare Diseases Are Now Chronic Diseases

Is Rare Disease a Unique Field of Medicine/Science?

- Conditions are typically genetic in nature
- Evidence based on small numbers of patients
 - Limited incidence data
 - Limited outcome data
- Patients/families are typically best source of information
- Typically seen by highly specialized caregivers
- Use multiple specialties with coordination for each dz.
- Care expertise is limited.
- Lifespan diseases

The Number of Rare Diseases with clear molecular diagnosis is exploding



Source: Online Mendelian Inheritance in Man, Morbid Anatomy of the Human Genome

How to Deal With The Depth and Width of Rare Diseases

- Networking
 - Isolation does not work
 - Everyone has to be at the table
- Natural History Studies (length vs depth)
- Sharing clinical practices
- Sharing diagnostic practices
- Flexibility
 - Different data models
 - Evolving standards
 - Current knowledge is already obsolete

How Do Rare Disease Clinical Research Networks Help

- The patients and care givers are the researchers
 - Questions gain relevance
 - Partnerships can be sustained over longer periods of time
- Collect longitudinal natural history data across multiple centers and long periods of time
- Clinical intervention strategies can be shared/compared
- Condenses the patients and caregivers in one system
 - Easier to do trials of new therapies
 - Easier for patients to find expertise

Programs In The USA

- NIH RDCRN (2004)
 - 22 current consortiums/databases
 - 197 Diseases
 - 454 Clinical Centers
 - 45,066 participants (as of Monday)
 - 21 countries
- The NORD Natural History Patient Registry Program (2013)
 - 26 Registry/Natural History databases
 - Data is owned by the patient organizations
 - Standardized software
 - Standardized ontologies, designed with U.S. FDA
 - Can partner with researchers, industry, other orgs.



NORD[®]
National Organization
for Rare Disorders

We Need and Must Develop Globally

- Common ethical standards
- Common Ontology/Descriptive language
- Common Platforms
- Evidence Standards for Rare Diseases
- Clinical Trial Standards for Rare Diseases
- NO field is better suited to make use of digital and informatics technology