

## **Day one-Plenary session B**

### **Alternatives and Future Promises for a Collaborative Rare Disease Registry**

**Chris Forrest M.D., Ph.D.**

This session will examine alternative approaches for the structure and organization of a collaborative rare disease patient registry. The talk will make the case that there is a new and growing imperative for a collaborative rare disease registry that supports research and healthcare improvement, articulate the necessary elements for success and sustainability, and options for collaboration presented as a staged model. The talk will also provide a vision of a future health information technology system that automatically populates the registry with a mix of electronic health record, personal health record, and research data, including biological specimen repositories, and enables collaboration between patients and scientists in a co-participatory research process. This bold vision will require an equally bold and thoughtful business model, which is the concluding topic.



*Uniting Rare Diseases*

*Advancing Rare Disease Research:  
The Intersection of Patient Registries, Biospecimen Repositories  
and Clinical Data*

*Plenary session B*

*Christopher B. Forrest*

*Mary D. Ames Professor of Pediatrics  
Children's Hospital of Philadelphia  
University of Pennsylvania School of Medicine*

*Structure & Function of a Collaborative Rare Disease  
Patient Registry*

# This Talk Will Address:

- ① Why now?
- ② How will this succeed?
- ③ “Stages of Collaboration”
- ④ Future Health IT Infrastructure
- ⑤ Business models

**(1) Why Now?**

# Key Element Necessary for Collaboration

## Shared Scientific Focus

Need to define “what is in it  
for me.”

# Why Collaborate?

1. Access for all patients with rare diseases
2. Share resources
  - Administrative
  - Technical
  - Human
3. Shared learning

# The “New” Science

- **Holistic:** nature as integrated whole
- **Network:** central metaphor, systems, knowledge is a network
- **Non-linear:** world is not a linear model, cannot be understood reductionistically
- **Generalism:** integrative, synthetic
- **Values:** cooperation, partnership

**(2) How will this  
succeed?**

# Elements of Success

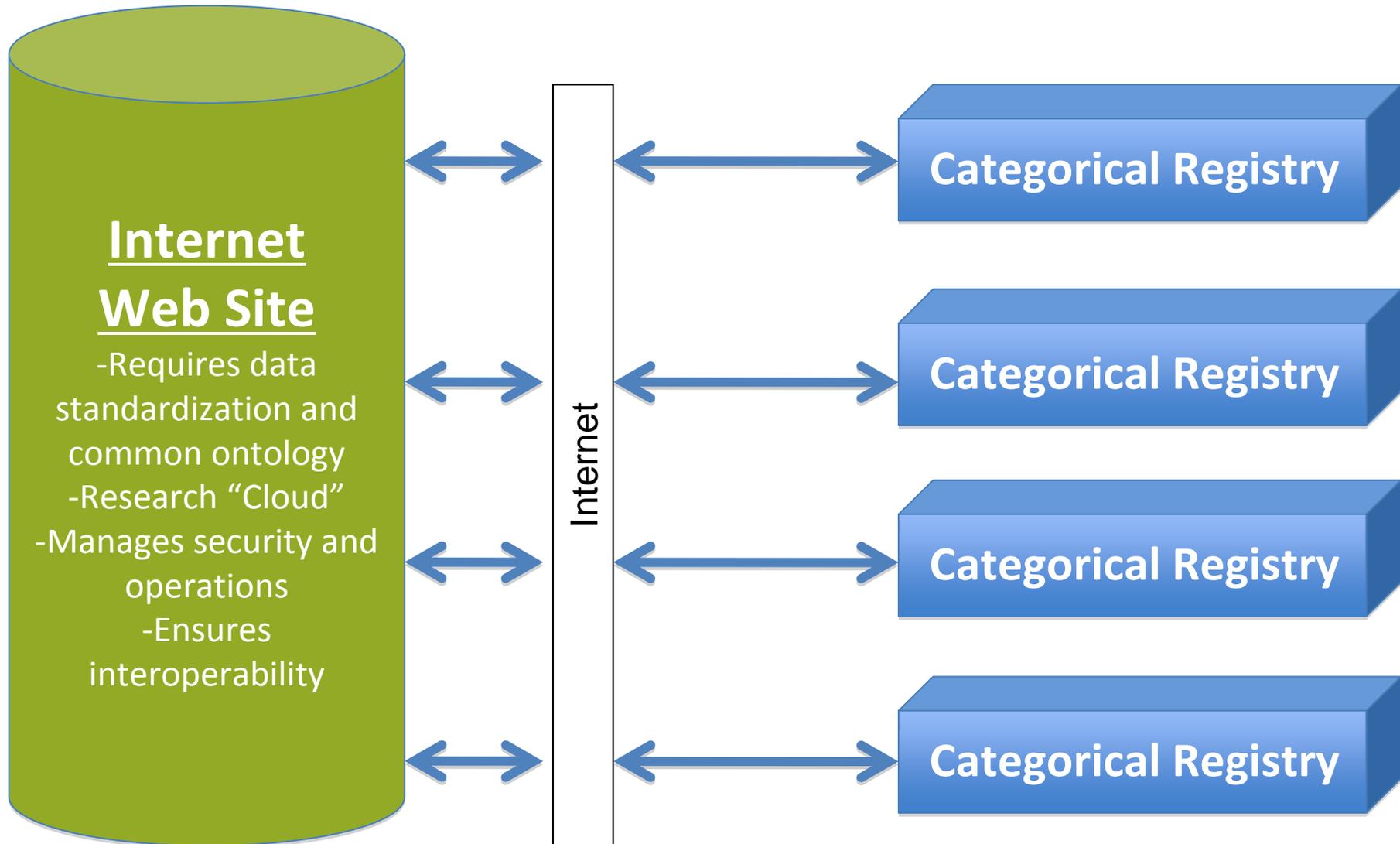
- **Shared vision (access for all; resource sharing; learning)**
- **Leadership**
- **Resources: time, money, intellectual**
- **Communication**
- **Trust and transparency**

# (3) Stages of Collaboration

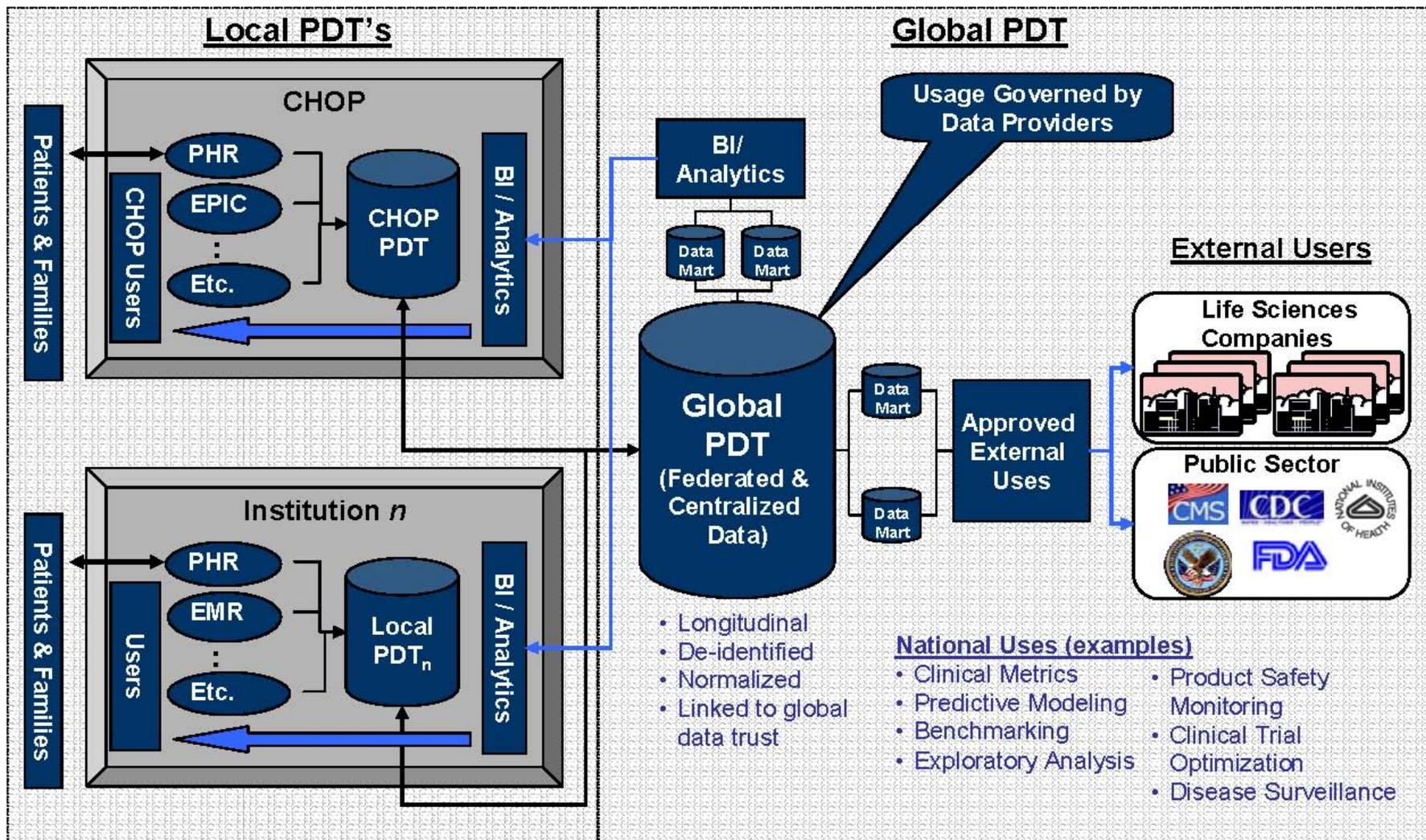
# Collaboration Stages

- **Affiliate:** Lists, tags, ad hoc
- **Standardize:** definitions, vocabularies, methods
- **Data Sharing**
  - Distributive: “data holders” retain data, queries, local autonomy, distributed governance
  - Federated: common database, cores, local and federated network projects

# Distributed Data Sharing Model



# Federated Data Model



# (4) Future Health IT Infrastructure

# The Future Health Information System

Shared Data Trusts
Knowledge Delivery Tools (Visual Data Mining, Decision Support)
Local Data Trusts
Population-Level Data
Molecular Diagnostics (-omics, Biomarkers)
Automated Capture of Patient Activity or Health Status (Biomonitors)
Patient-Reported Information (PHR)
Electronic Clinical Documentation (EMR)
Curated Clinical Data

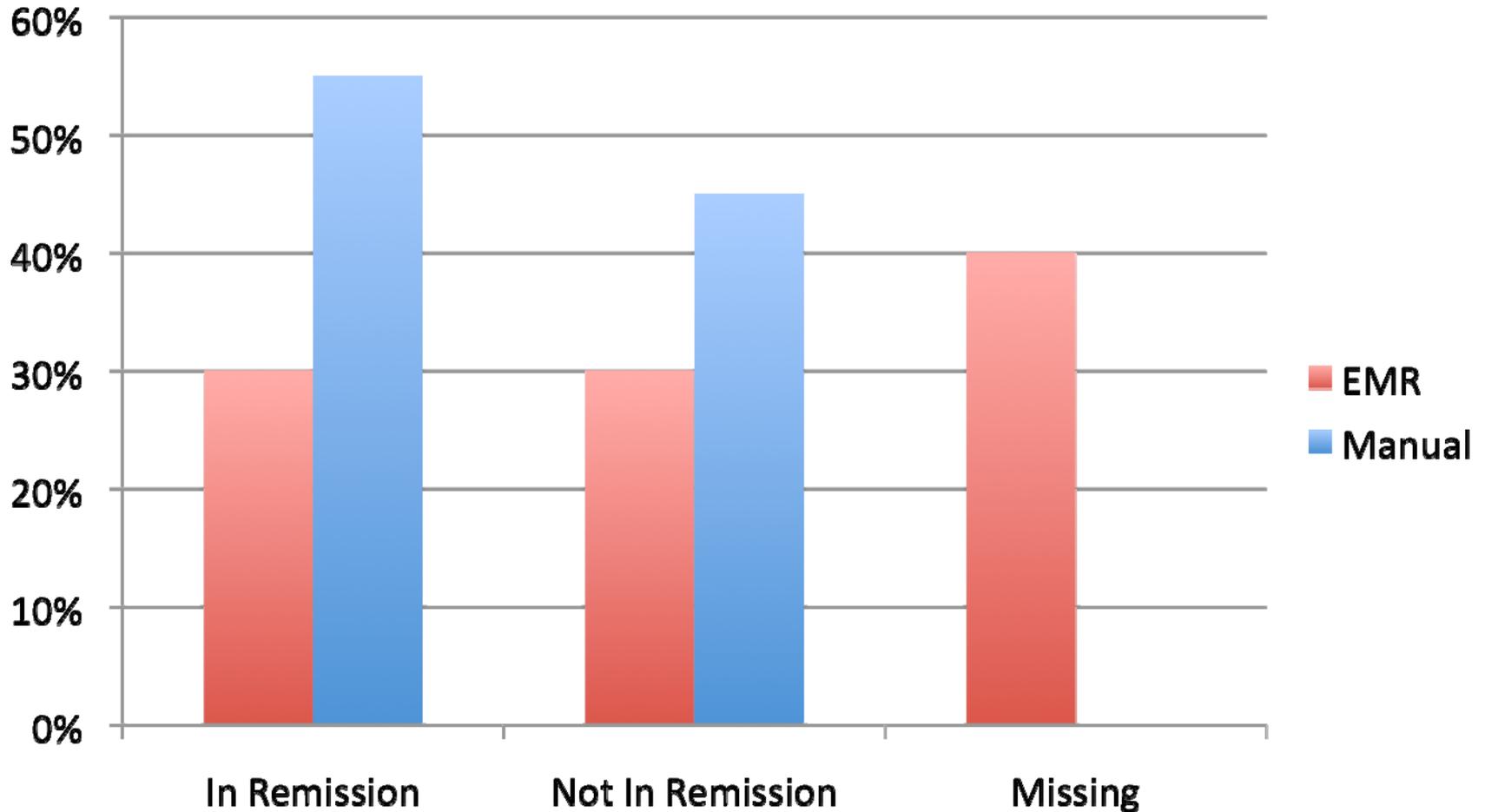
## EMR Data Quality

# Juvenile Idiopathic Arthritis Remission Definition

- No “active” joints
- No enthesitis
- No uveitis
- No systemic symptoms
- < 15 minutes of morning stiffness

If all 5 criteria are met, the patient is in remission (Primary Outcome)

# JIA Disease Remission: Manual/Curated versus EMR Data



# In 1 day at CHOP, clinicians recorded in the EMR . . .

- 278 ways to describe fever for 465 patients
- 123 different ways to express ear pain for 213 patients
- 34 different ways to describe h/o ear drainage for 83 patients
- 23 different ways to describe 45 ill appearing children
- 99 different ways to describe 284 red ears
- 444 different ways to describe 6,148 non-red ears

# **(5) Business Models**

# Business Models

- Need to consider both funding and governance

## Options

1. Public funding and governance
2. Academic health center supported and administered
3. Public funding / private governance