

Data Fusion in Phenotypes EHR, Registry, PRO

Ken Mandl

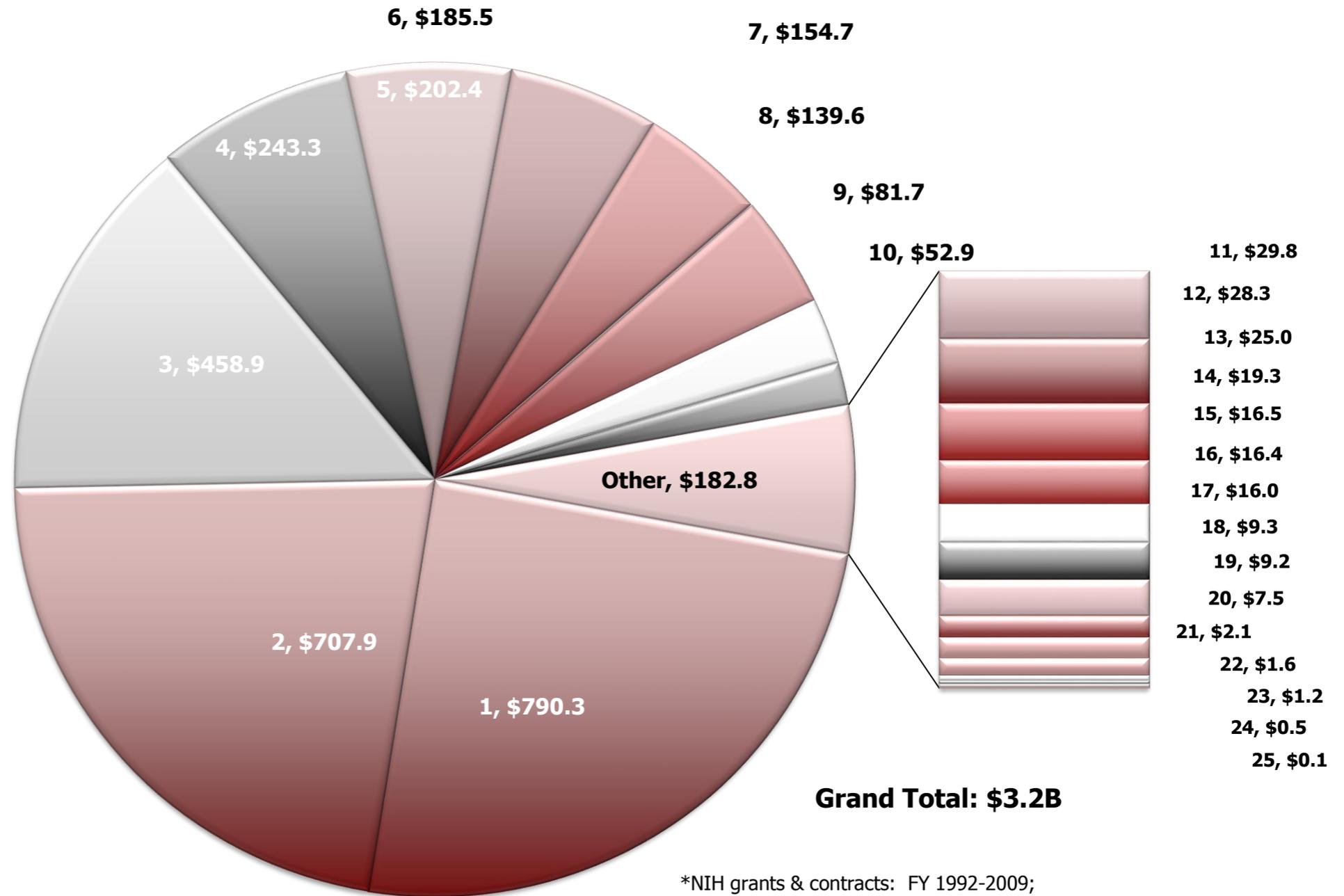
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ATSDR: Agency for Toxic Substances & Disease Registry
FIC: Fogarty International Center
NCBBB: Nat'l Center on Birth Defects & Developmental Disabilities
NCCDPDP: Nat'l Center for Chronic Disease Prevention & Health Promotion
NCI: National Cancer Institute
NCPDCID: Nat'l Center for Preparedness, Detection, & Control of Infectious Diseases
NCRR: Nat'l Center for Research Resources
NEI: National Eye Institute
NHLBI: Nat'l Heart, Lung, & Blood Institute
NIA: National Institute on Aging
NIAAA: Nat'l Inst on Alcohol Abuse & Alcoholism
NIAD: Nat'l Institute of Allergy & Infectious Diseases
NIAMS: Nat'l Institute of Arthritis & Musculoskeletal & Skin Diseases
NIBIB: Nat'l Institute of Biomedical Imaging & Bioengineering
NICHD: Nat'l Institute of Child Health & Human Development
NIDA: Nat'l Institute on Drug Abuse
NIDCD: Nat'l Institute on Deafness & Other Communication Disorders
NIDCR: Nat'l Institute of Dental & Craniofacial Research
NIDDK: Nat'l Institute of Diabetes & Digestive & Kidney Diseases
NIHES: Nat'l Institute of Environmental Health Sciences
NIGMS: Nat'l Institute of General Medical Sciences
NIMH: Nat'l Institute of Mental Health
NINDS: Nat'l Institute of Neurological Disorders & Stroke
NIOSH: Nat'l Institute for Occupational Safety & Health
NLM: National Library of Medicine

Fig. 2: Cumulative Registry Funding* for FY 2009 Active Projects by Institute or Center (in millions, FY 2008 CPI adjusted dollars)



*NIH grants & contracts: FY 1992-2009;
 CDC grants & contracts: FY 1995-2009

More Common in Non-Registry

ICD Text	Absolute*/Relative Proportion
CONGEN MITRAL STENOSIS	13.5*
RESPIRATORY DISTRESS SYN	11.4*
AWAIT ORGAN TRANSPLNT ST	9.0*
ENDOCARD CUSHION DEF NEC	8.1*
MECONIUM ASP W RESP SYMP	7.4*
CONG AORTA VALV STENOSIS	7.4*
HYPOPLAS LEFT HEART SYND	7.2*
CARDIAC ARREST	7.2*
NEONATAL THROMBOCYTOPEN	7.0*
AORTIC ATRESIA/STENOSIS	6.9*
HEART VALVE REPLAC NEC	6.5*
OSERV-SUSPECT INFEC INFNT	6.3*
NEONATAL DEHYDRATION	6.3*
NB TRANSITORY TACHYPNEA	6.1*

ICD Text	Absolute*/Relative Proportion
CORONARY ARTERY ANOMALY	5.8*
ATRIOVENT BLOCK-1ST DEGR	5.8*
HEART TRANSPLANT STATUS	5.6*
Hypoxc-ischem enceph NOS	5.4*
MITRAL VALVE DISORDER	2.3
CONGESTIVE HEART FAILURE	1.9
UNSPECIFIED DIASTOLIC HEART FAILURE	1.8
AORTIC VALVE DISORDER	1.8
COARCTATION OF AORTA	1.7
CONG SUBAORTIC STENOSIS	1.5
DOWN-s SYNDROME	1.4
CONG PULMON VALVE STENOS	1.3
CONG AORTA VALV INSUFFIC	1.3

* 0% prevalence among registry patient; number shown is absolute percent of non-registry patients

More Common in Registry

ICD Text	Absolute*/Relative Proportion	ICD Text	Absolute*/Relative Proportion
PSYCHOGEN CARDIOVASC DIS	18.2*	DELAYED MILESTONE	0.3
CHR AIRWAY OBSTRUCT NEC	10.2*	AGENESIS OF LUNG	0.4
RETINOPH PREMATURITY NOS	10.2*	UNSPECIFIED WEEKS OF GESTATION	0.4
Hx-cong malform-digest	9.7*	RESP SYSTEM DISEASE NOS	0.5
GLUCOCORTICOID DEFICIENT	6.8*	PULMONARY ARTERY ANOM	0.5
LBW 500-999 GRAMS	6.3*	PERINATAL CHR RESP DIS	0.5
LUNG TRANSPLANT STATUS	6.3*	MULT CONGEN ANOMAL NEC	0.5
TRACHEOST COMPL OTH	6.3*	CARDIAC DYSRHYTHMIA NOS	0.5
MACHINE DEPENDENCE NOS	5.7*	DEPEND ON MACHINES/SUPPL OXYGEN	0.5
PULMONARY VALVE ANOM NEC	5.1*	LOW BIRTH WEIGHT UNSPEC	0.5
DIAPHRAGMATIC HERNIA	0.2	HISTORY OF ECMO	0.5
ANOMALIES OF DIAPHRAGM	0.3	PULMONARY VALVE DISORDER	0.5
OBS-SUSP CARDIOVASC DIS	0.3	≥ 37 COMPLETED WEEKS OF GEST	0.6
CARDIAC MURMURS NEC	0.3	TRACHEOSTOMY STATUS	0.6
OBSTRUCTIVE SLEEP APNEA	0.3		

* 0% prevalence among non-registry patient; number shown is absolute percent of registry patients



C3-PRO

RESEARCH ARTICLE

C3-PRO: Connecting ResearchKit to the Health System Using i2b2 and FHIR

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Abstract

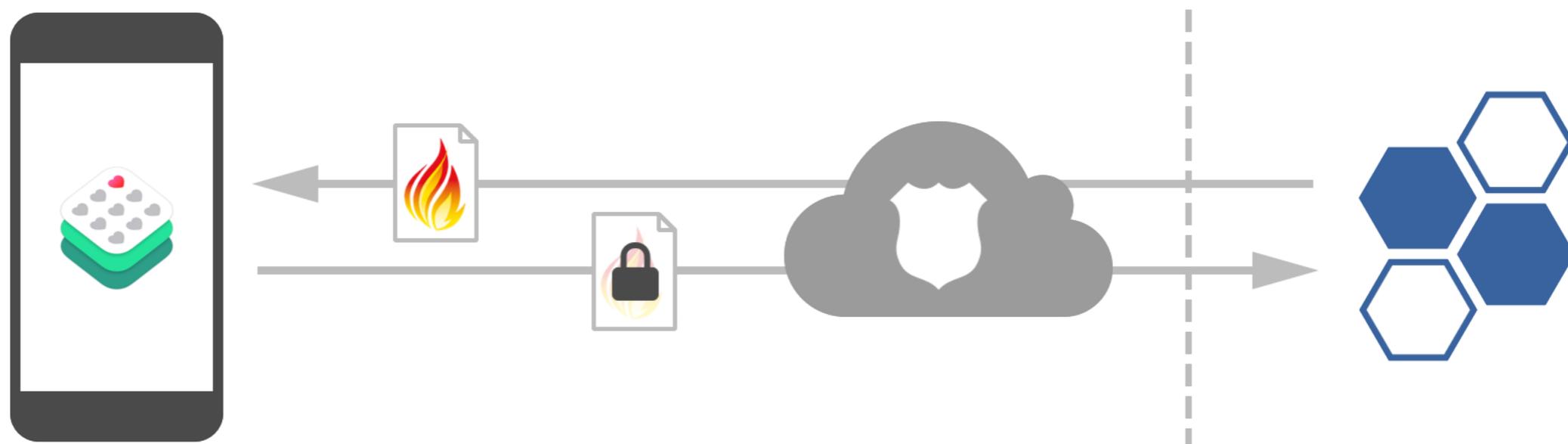
A renewed interest by consumer information technology giants in the healthcare domain is focused on transforming smartphones into personal health data storage devices. With the introduction of the open source *ResearchKit*, Apple provides a framework for researchers to

 OPEN ACCESS

ResearchKit

- Open Source iOS Framework by Apple
- Guides through consent
- Survey rendering
- Multitude of “tasks”
 - Tapping speed
 - 6-min walk distance
 - memory game & more





C3-PRO

C3-PRO

Consent, Contact and Community framework for PRO

- Mobile interface: [ResearchKit](#)
- Content and wire format: [FHIR](#)
Fast Healthcare Interoperability Resources
- “Receiver server” publicly hosted
- Research backend: [i2b2](#)

Enrolling Users

- FHIR Contract resource
- Ask eligibility questions
- Guide through consent
- Collect name and signature, create PDF on device
- HIPAA “Safe Harbor” compliant patient data, identified by random UUID, sent as Patient resource, signed Contract resource

Surveys and Activity Data

- Longer surveys every 4 months, shorter ones every 2 weeks
- Questions about antiviral medication therapies and prescription patterns; insurance status
- *SF-36 Physical Functioning Scale* and *WPAI:Hepatitis C* questionnaires
- Activity data: minutes spent sleeping/sitting/exercising, steps taken, flights climbed

Surveys

- Download Questionnaire FHIR resource, fallback to cached version
- Translate into ResearchKit surveys
- Convert answers into QuestionnaireResponse FHIR resource
- Encrypt and send to C3-PRO receiver

ResearchStack

- Porting C3-PRO over with Deborah Estrin

<http://c3-pro.chip.org/>

- C3-PRO
 - Marc Natter
 - Pascal Pfiffner
 - Isaac Pinol Catadau
- SCILHS
 - Shawn Murphy
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