All of Us RESEARCH PROGRAM

Application of a Data Quality Framework to Ductal Carcinoma In Situ Using Electronic Health Record Data From the All of Us Research Program

OHDSI Community Call

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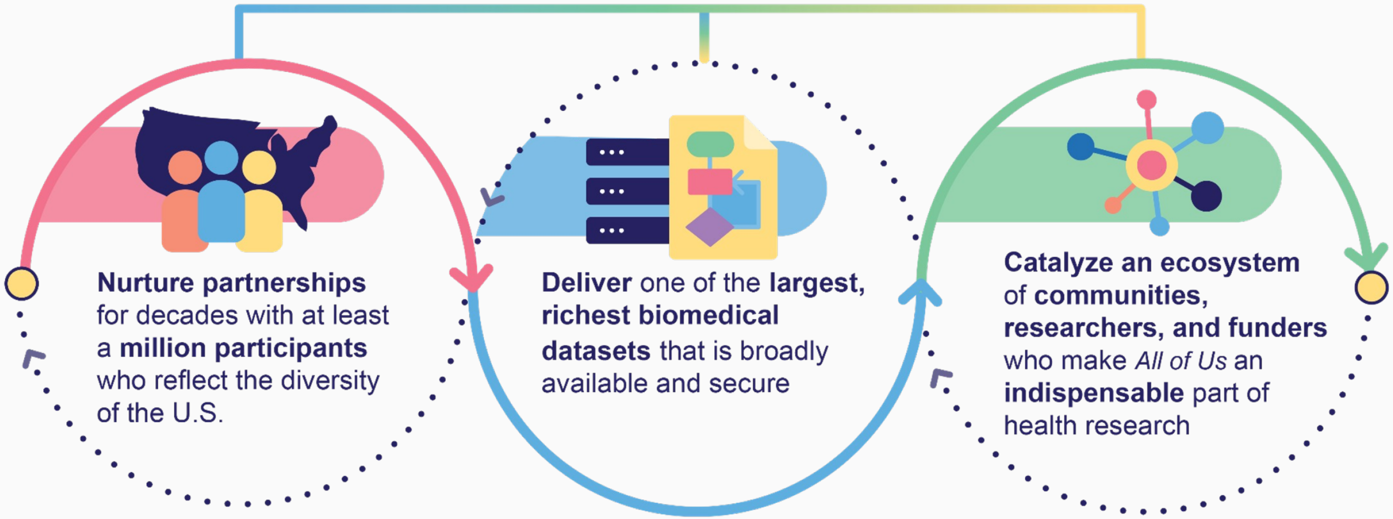
All of Us Research Program

NIH National Institutes of Health

The *All of Us* Research Program Mission

Accelerate health research and medical breakthroughs, enabling individualized prevention, treatment, and care for all of us



Nurture partnerships for decades with at least a **million participants** who reflect the diversity of the U.S.

Deliver one of the **largest, richest biomedical datasets** that is broadly available and secure

Catalyze an ecosystem of **communities, researchers, and funders** who make *All of Us* an **indispensable** part of health research

Made possible by a team that maintains a culture built around the program's core values

Study Objectives

Specific Aims

- Develop and operationalize an electronic health record (EHR) data quality framework
- Apply the dimensions of the framework to the phenotype and treatment pathways of ductal carcinoma in situ (DCIS) using *All of Us* Research Program data
- Propose and apply a checklist to evaluate the framework's application

Why is it significant?

- Provides insights into the fitness of using *All of Us* EHR data for specific phenotypes
- Understand the strengths & weaknesses of checklists for other phenotypes

Ductal Carcinoma In Situ Public Health Impact

- DCIS is a precancerous condition that accounts for about 25.0% of breast cancers diagnosed in the United States [1, 2]
- DCIS is a well-known and understood disease, has a clearly defined treatment protocol and, if treated, shows excellent disease-free survival without additional surgery [3]
- The incidence of DCIS has increased in recent years due to the widespread use of screening mammography [1]

[1] Allegra CJ, Aberle DR, Ganschow P, et al,. National Institutes of Health State-of-the-Science Conference Statement: Diagnosis and Management of Ductal Carcinoma In Situ September 22–24, 2009. J Natl Cancer Inst. 2010; 102(3):161–169.

[2] Sarah E Pinder and Ian O Ellis. Review:The diagnosis and management of pre-invasive breast disease Ductal carcinoma in situ (DCIS) and atypical ductal hyperplasia (ADH) — current definitions and classification. Breast Cancer Res 2003, 5:254-257

[3] About Clinical Practice Guidelines. National Comprehensive Cancer Network. 2023. Accessed October 30, 2023. https://www.nccn.org/guidelines/guidelines-process/about-nccn-clinical-practice-guidelines

Methods: Study Design

- **Phenotype**
  - ***Cases:*** Earliest occurrence of any ICD-09/10 code or SNOMED concept for DCIS mapped to OMOP concepts codes and restricting to female participants who were at least 18 years of age
  - ***Controls:*** Female participants who were at least 18 years of age and did not have a DCIS diagnosis

- **Clinical Measures and Interventions:** National Comprehensive Cancer Network (NCCN) DCIS related treatment guidelines (workup, primary treatment, and postsurgical treatment)

- **Data quality framework:** conformance, completeness, concordance, plausibility, temporality

Definitions: Data Quality Dimensions

- **Conformance:** Dataset values and elements (standards, syntax, and structure) were equivalent or represented in the same way

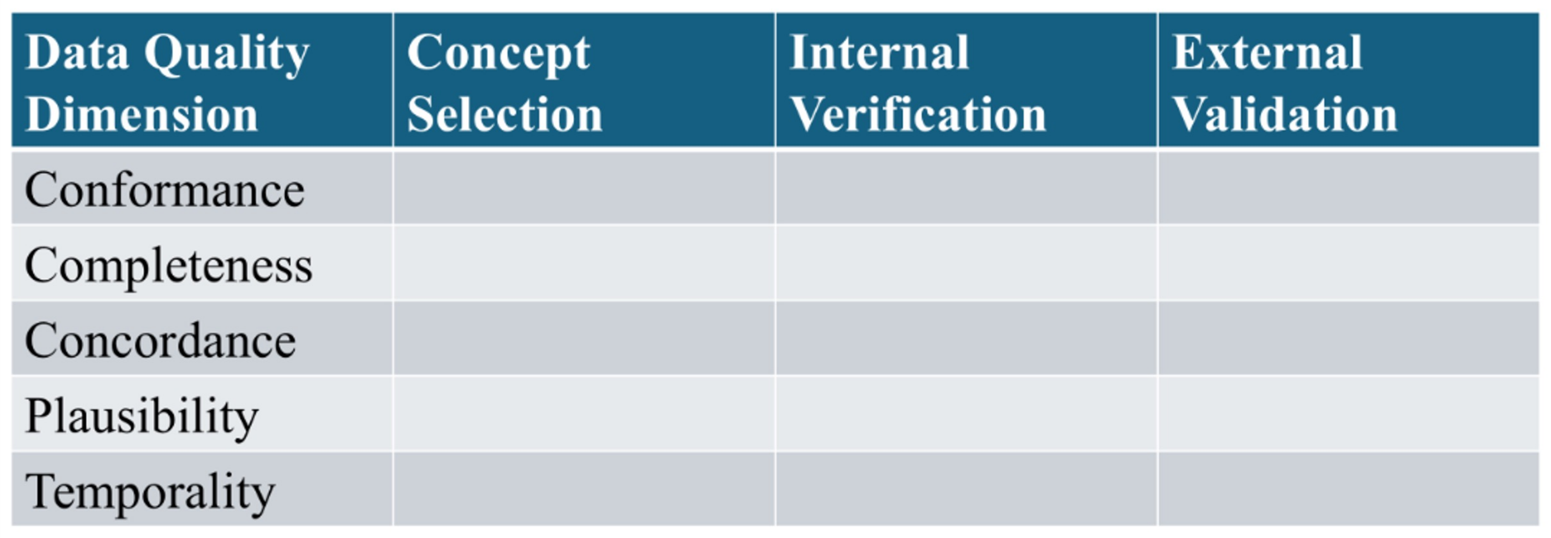
- **Completeness:** Dataset values and elements have been captured/were available

- **Concordance:** Dataset values and elements were similar or in agreement

- **Plausibility:** Dataset values and elements were believable

- **Temporality:** Dataset values and elements had valid start times, end times, and durations and followed expected order

Methods: Data Quality Dimensions Evaluability Checklist



Data Quality Dimension	Concept Selection	Internal Verification	External Validation
Conformance			
Completeness			
Concordance			
Plausibility			
Temporality			

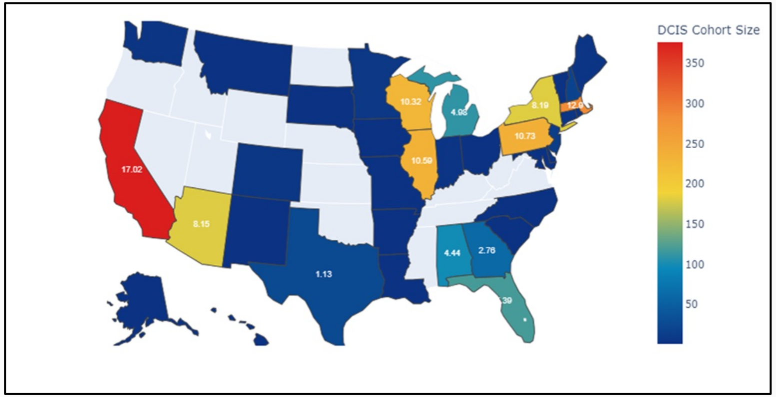
Source: Berman L, Ostchega Y, Giannini J, et. al. Application of a Data Quality Framework to Ductal Carcinoma in Situ Using Electronic Health Record Data from the All of Us Research Program. JCO Clinical Cancer Informatics.2024 Aug:8:e2400052.

DCIS Cohort Demographics and Geographic Distribution

- Out of 365,488 participants in *All of Us* more than 350,000 have shared their EHR. Among these participants, 2,209 are females with DCIS (0.6%)

- In the *All of Us* DCIS cohort, 52.5% of the females were diagnosed between the ages of 60-79, and 66.0% of the diagnosed population is non-Hispanic White

- The highest percentages of the *All of Us* DCIS cohort are in California (17.0%), Massachusetts (12.0%), Pennsylvania (10.7%), and Illinois (10.5%)

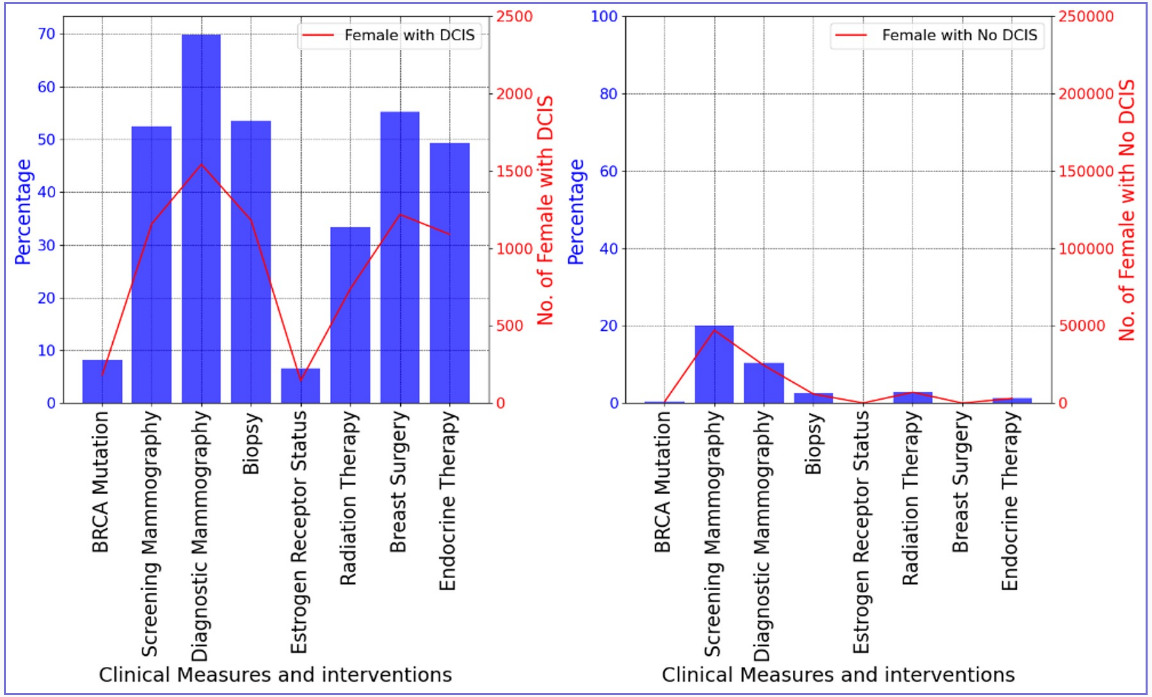


Conformance

Assessing the source distributions of the OMOP concept codes

- ICD 9/10 only: 1,924 (87.1%)
- ICD 9/10 and SNOMED: 277 (12.5%)
- SNOMED only <= 20

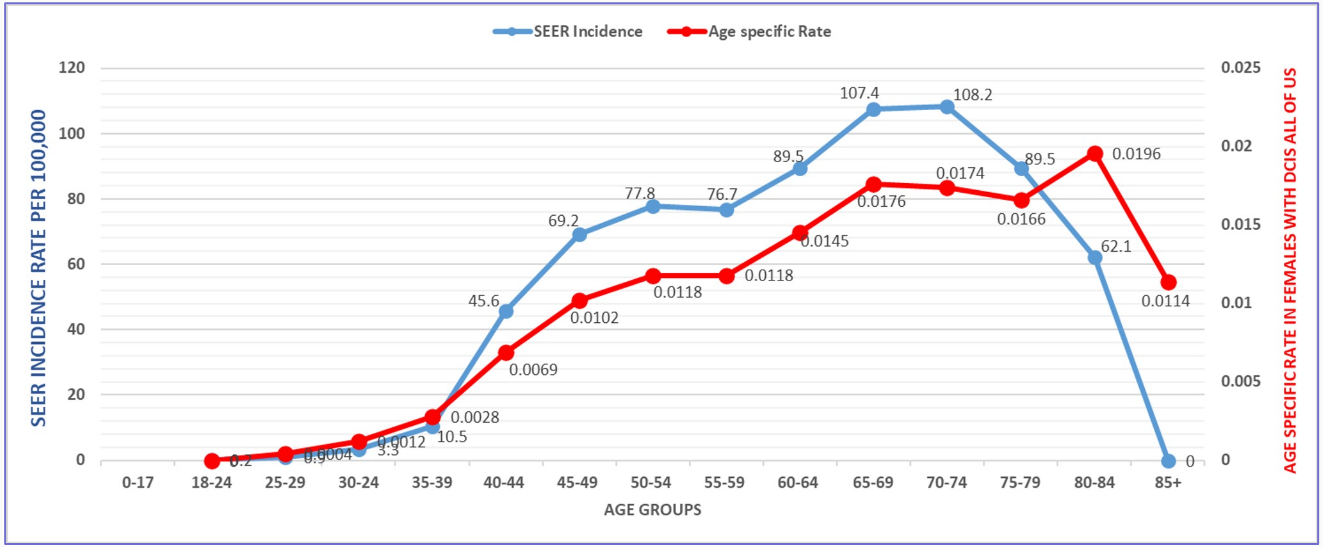
Completeness



The DCIS and non-DCIS groups showed differences in the proportions of NCCN guideline related concepts, which included diagnostic mammography (69.9% vs. 11.2%), biopsy (53.5% vs. 3.2%), surgery (55.18% vs. 1.3%), and endocrine therapy (49.4% vs. 1.9%), ($p<0.01$).

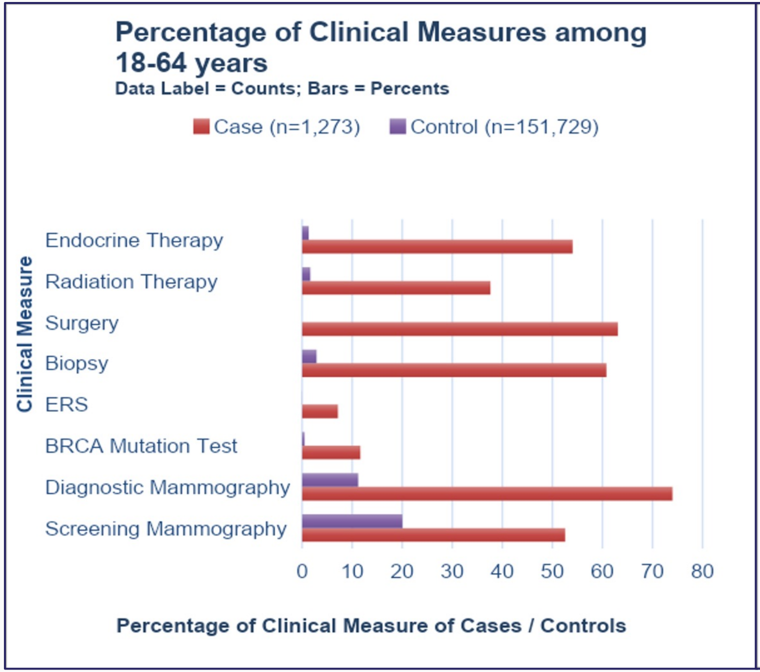
Concordance

The correlation between DCIS *All of Us* age-specific prevalence rates and SEER (1975-2017) reported incidence of DCIS by age groups



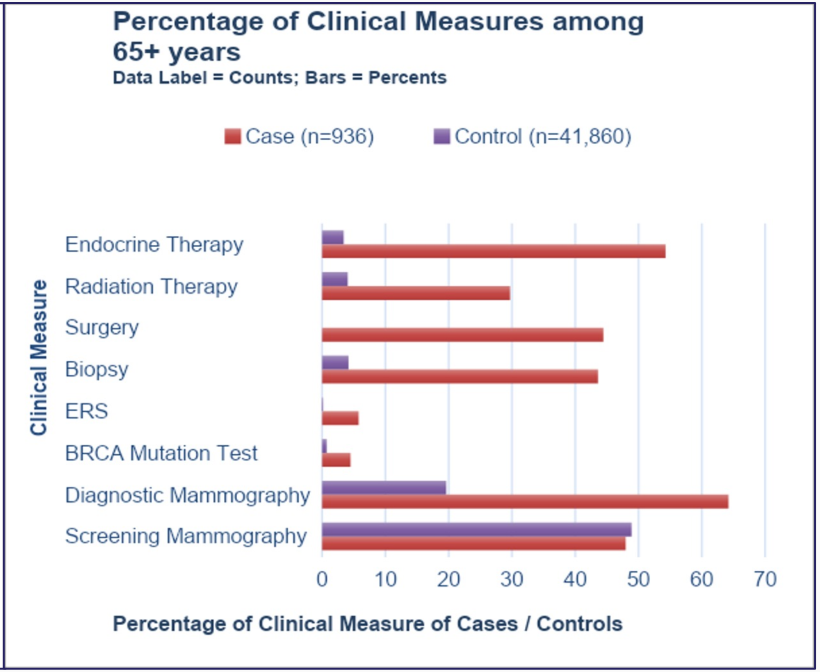
The Spearman rank correlation coefficient between these two sets of values is 0.85 (p<0.01).

Plausibility



Percentage of Clinical Measures among 18-64 years

Data Label = Counts; Bars = Percents



Percentage of Clinical Measures among 65+ years

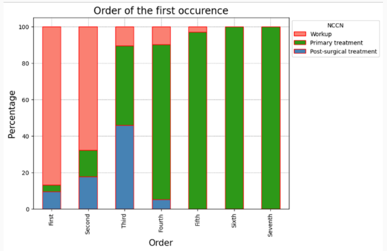
Data Label = Counts; Bars = Percents

The relative number of cases and controls who had clinical measures and interventions differed by age group p<0.01.

Temporality

- Biopsy-Diagnosis Interval
  - Antecedent biopsy data were available for 1,023 females (47%)
  - The median time from biopsy to diagnosis was 8 weeks

- NCCN Phase Analysis
  - Organized clinical measure and intervention concept sets by NCCN phase guidelines (e.g. workup, primary treatment, postsurgical treatment)
  - Sequenced the concept sets by those phases
  - Most participants progressed from workup to primary treatment



Key Findings

- Developed a systematic and generalizable approach to assessing phenotype data quality

- All five dimensions were evaluated successfully for concept selection, internal verification, and external validation. External validation was limited by external benchmarks

- Used case and control phenotype definitions to ensure there are differences between these groups

Limitations and Next Steps

- Limitations
  - Over-representation of academic medical centers / healthcare provider organizations
  - Manual selection of concepts limited by knowledge of clinical experts
  - Fragmented EHR data
  - Absence of data from unstructured sources

- Next Steps
  - Application of the framework to other phenotypes
  - Refinement of temporal analysis

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