OHDSI Rare Diseases Working Group

2025 OKR

WG leads:
Xiaoyan Wang PhD
Chunhua Weng PhD

Rare Diseases Working Group

Mission Statement:

To advance the understanding, diagnosis, and treatment of rare diseases through the development and application of open-source analytics tools, standardized data models, and collaborative research.

- Develop and refine methodologies for collaborative rare disease case aggregation within the OHDSI community.
- Support evidence generation over rare diseases within the OHDSI ecosystem.
- Facilitate collaboration across stakeholders, including researchers, clinicians, sponsors, industry partners, and patient advocacy groups, to drive impactful discoveries.

Rare Disease WG 2025 OKRs

WG Leads: Xiaoyan Wang, Chunhua Weng

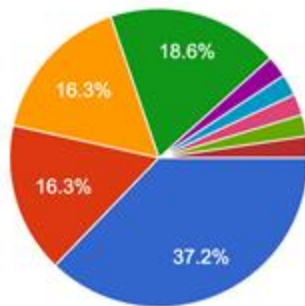**Objective 1**: **Strengthen community engagement and collaboration**

KR1: Form a collaborative team of ~20 active members with expertise in rare disease research, data science, phenotyping and clinical study design.

KR2: Organize about three knowledge-sharing sessions to foster interdisciplinary collaboration.

KR3: Establish partnerships with key stakeholders, including researchers, clinicians, patient advocacy groups, regulatory agencies, and industry leaders, to enhance research impact.
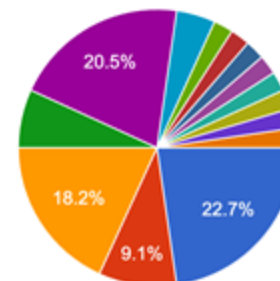

Please indicate your primary affiliation (choose one):
43 responses

- Academic/Research
- Pharmaceutical/Biotech
- Technology/Software
- Consulting/CRO
- Healthcare Provider
- Patient Advocacy Group
- Driving change and adoption of OMOP in Sweden. Engaged in an IHI Call 9 i...
- Health Authority representative (MD)
- Nonprofit Research and Data Coordin...

18.6%
16.3%
16.3%
37.2%

What expertise, skills, or data assets can you contribute to the working group? (Chose one option that fits best)
44 responses

- Clinical and Real-world data (RWD)
- Rare disease research
- Data standardization
- Patient engagement/advocacy
- Statistics, Analytics, AI/ML application...
- Clinical trial design and Pharmacoepid...
- Regulatory science
- Change management, Analytics, Lead...

20.5%
18.2%
9.1%
22.7%

1/2

Rare Disease WG 2025 OKR

WG Lead: Xiaoyan Wang, Chunhua Weng

- **Objective 2 : Enable scalable and reproducible rare disease analytics**

  - **KR1**: Design and initiate a multi-site rare disease study leveraging OHDSI's network and the OMOP CDM.

  - **KR2:** Develop a standardized approach for identifying and integrating hard-to-find rare disease cohorts across multiple data sources.

  - **KR3:** Release preliminary findings and methodological insights to guide future large-scale rare disease research.

- **Objective 3: Integrate Innovative AI Technologies for Rare Disease Research**

  - **KR1:** Evaluate and implement NLP and LLMs for rare disease cohort identification.

  - **KR2:** Develop and test the integration of knowledge graphs and AI-driven knowledge discovery tools for rare disease evidence generation.

  - **KR3:** Release an open-source framework or best practices document for leveraging AI in rare disease studies within the OHDSI community.

Rare Diseases WG Next Meeting

- To join, go to ohdsi.org -> workgroups tab ->
- Select Workgroup - Rare Diseases
- Meeting details:

    Time: 10-11am EST, Feb 28th, 2025

    Meeting Link: Join the meeting now

    Meeting ID: 210 677 103 238

    Passcode: Yw2GJ7pq

    Agenda:
    – Introduce the WG's objectives
    – Discuss priority areas
    – Identify opportunities for collaboration.