OHDSI Rare Diseases Working Group

Feb 2026

WG leads:
Xiaoyan Wang PhD
Chunhua Weng PhD

Rare Disease Working Group Sessions

- **Foundations of Real-World Evidence Generation**
- *Important Groundwork for Real-World Evidence Generation in Pulmonary Arterial Hypertension (PAH)*
  Speaker: Eva-Maria Didden, MD, Johnson & Johnson
- **Rare Disease Terminologies & Vocabularies**
- *Integration of Rare Disease Terminologies into Rare Disease Research*
  Speaker: Bryan Laraway, University of North Carolina at Chapel Hill
- *Rare Disease Vocabularies: Challenges and Solutions*
  Speakers:
  Melissa Haendel, PhD, University of North Carolina at Chapel Hill; Amol Bhalla, PhD; Kerri Grizer (IMO)
  Alexander Davydov, PhD (OHDSI Vocabulary Team)
- *Orphanet Nomenclature: The Standard Medical Terminology for Rare Disease Coding*
  Speakers: Caterina Lucano, PhD; Ana Rath, MD, PhD, Orphanet
- **Data Quality, Standards, and Collaboration**
- *Data Partnerships and Quality Standards for Collaborative Research in the OHDSI Community*
  Speaker: Clair Blacketer, MPH, Director, Johnson & Johnson
- **Clinical Applications & Feasibility Studies**
- *Early Detection of CHAPLE Disease in Pediatric Protein-Losing Enteropathy: A Feasibility Study*
  Speaker: Kyeryoung Lee, PhD
- *Maximizing EHR Semantic Meaning for Rare Diseases Using a Direct Mapping Strategy*
  Speaker: Melanie Philofsky, MS, RN, Director of Clinical Informatics

Rare Diseases Working Group Symposium 2025

- **Invited Deep Diving Sessions**

Michele Zoch, PhD – Institute for Medical Informatics and Biometry, TU Dresden (Germany)

Topic: Integrating ORPHAcodes into ATHENA and research directions in Klinefelter syndrome & McCune– Albright syndrome

Zhandong Liu, PhD – Associate Professor, Pediatrics & Neurology, Baylor College of Medicine (USA)

Topic: Rare disease diagnosis using biomarkers

Radek Wasiak, PhD – rAIre Rare Disease Insights (EU)

Topic: The evolving role of real world data in rare disease drug approval and reimbursement - from methods to new data sources

- **Globalization of RWE in Rare Diseases: challenges/strategies**

Rare Disease WG 2025 OKRs

- **Objective 1: Community engagement and collaboration**
  - KR1: Continue the collaborative team of ~20 active members with expertise in rare disease research, data science, phenotyping and clinical study design.
  - KR2: Organize monthly 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.
- **Objective 2: Exploring scalable rare disease analytics**
  - KR1: Design and initiate a multi-site rare disease study leveraging OHDSI's network and the OMOP CDM.
  - KR2: Develop a in-depth approach for identifying and integrating hard-to-find rare disease cohorts across multiple data sources.
    - Structured data standardization
    - Deep clinical granularity
  - KR3: Release preliminary findings and methodological insights to guide future large-scale rare disease research.