Rare Disease Community - Meeting Resources					
Topic	**Description**	**Date & Time**	**Lead**	**Slides**	**Recording**
Data Collection for Rare Disease	The meeting focused on discussing tools for head-to-toe phenotyping in rare disease research. Zohreh presented RareX, a research platform of Global Genes that facilitates symptom-level data collection for rare diseases through structured surveys based on yes/no responses across body systems. Ada then demonstrated PhenoDB, a clinician-focused tool with 3,900 terms mapped to HPO that allows comprehensive phenotyping with 22 upper-level organ system categories, including both structural and functional assessments. The discussion covered key challenges including ontology mapping for terms with multiple examples, translation requirements for global implementation, and the need for better engagement from different stakeholders including patients, caregivers, and clinicians. Participants raised important	2026-05-27	Zohreh Talebizadeh and Ada Hamosh		Recording Passcode: GkDFBf%7
Rare Disease Phenotyping Challenges	The Rare Disease (RD) Community of Interest (CoI) meeting focused on improving rare disease phenotyping approaches so they are both clinically meaningful and computationally actionable. Participants discussed how current standards — including phenopackets, Human Phenotype Ontology (HPO), and Observational Medical Outcomes Partnership (OMOP) — often "flatten" clinical narratives, making it difficult to capture important contextual details such as time course, comorbidities, pertinent negatives, prior history, environment, and access to care. The group emphasized that effective phenotyping models must balance structured, scalable data with the richness of clinical narratives to better reflect real-world clinical reasoning.	2026-02-24	Ada Hamosh		Recording Passcode: nW52%.#.L
HiFi Solves: a global federated network to accelerate collaborative RD research	HiFi Solves is a federated data network connecting renowned research institutions worldwide to build a real-time, globally representative database of rare genomic variants identified by HiFi long-read sequencing. Through this unique collaboration, HiFi Solves is accelerating innovation in the study of rare diseases and specific regions of the genome.	2025-06-04	Marc Fiume	Rare Disease Community Mee	Recording
Towards collaborative, decentralised ontology management for rare disease	Rare diseases are among the most complex medical challenges and undeniably require global coordination. Terminologies and ontologies act as hubs, connecting information generated by a wide range of often isolated resources, each containing crucial insights about individual diseases. Many of these resources use their own terminologies or standard vocabularies to describe rare disease data, making the development of an integrated "ontology of terminologies" all the more vital. However, managing such a resource is challenging and demands the integration of diverse communities and perspectives on rare diseases. The Mondo team rises to this challenge through community curation, incorporation of externally managed content, and the management of rare disease designations.	2025-05-07	Nico Matentzoglu	Rare Disease Community Mee	Recording
TISLab - Layperson Phenotyping	The presentation highlighted how empowering patients to contribute structured, layperson-friendly phenotype data—using more advanced, graph-based models beyond traditional coding systems—can improve diagnostic accuracy, especially in rare diseases, and help build a more effective, data-driven healthcare system.	2025-03-05	Julie McMurry	GA4GH RD Phenotyping 2025	
Advancing Rare Disease Knowledge Through Global Collaboration Webinar	Webinar on February 27, 2025 to celebrate Rare Disease Day. Hosted by the GA4GH Rare Disease Community of Interest, the webinar focused on collaborative efforts to advance rare disease knowledge and enhance global strategies for patient engagement.	2025-02-27	Daria Julkowska, Charl	Rare Disease Day 2025 - Web	Advancing Rare Disease Knowled
Phenotips	The session showcased how Phenotips enhances rare disease diagnosis by integrating structured phenotyping, AI-driven HPO extraction, and clinical workflows with EHR systems, while supporting data sharing and collaborative initiatives like Matchmaker Exchange and undiagnosed disease hackathons.	2025-02-05	Orion Buske	GA4GH RD CoI - PhenoTips - 2	
DECIPHER	Sharing rare disease phenotype-linked variant data for diagnosis and research	2024-08-07	Mallory Freeberg	20240807-DECIPHER-RD-Comn	
Mondo Disease Ontology	Building a Community-driven Disease Resource	2024-06-26	Sabrina Toro	Mondo - GA4GH rare	
Grand Challenges in Rare Disease Webinar	Orphanet is a unique online resource gathering and improving a vast knowledge base on Rare Diseases, to improve the diagnosis, care, and treatment of patients with Rare Disease. Switching gears, Matthew Bolz-Johnson from EURODIS presented on the patient experience within the Rare Disease diagnosis journey. Individuals with rare conditions live at the intersection of multiple vulnerable groups, which together have an accumulative impact on poor mental health. Stressors associated with rare conditions include but are not limited to 1) the diagnostic odyssey, 2) living with uncertainty, 3) complex interacting emotional and physical symptoms, and 4) stress and strain on relationships. To help navigate stressors, psychosocial protection factors such as being a part of a connected community and having a positive support system have been shown to positively influence mental health. EURODIS is currently working on a position paper which calls for the implementation of programs to promote mental health and psychosocial support for individuals living with rare conditions.	2024-02-29	Marc Hanauer, Matthew	Rare Disease Day 2024 - Webi	
ClinGen curation of ClinVar		2024-01-31	Heidi Rehm	Sharing_Patient_Data_in_ClinVa	
MAxO		2024-01-31	Leigh Carmody	GA4GH_MAXO_2024Jan31.pptx	