

Orphan Diseases Working Group
2025 Autumn Conference

The orphan disease working group met in Amsterdam to discuss ongoing and future scientific work. The group welcomed several new members. We reviewed progress and contributions from various authors to the three manuscripts in progress (CGI in Rare Disease Trials, Measuring Cognition in Rare Disease, Ethical Issues in Rare Disease). The CGI and Cognition manuscripts have interim drafts but work has been paused due to the government shutdown. Work will be commenced in December, and timelines for the papers shifted by one quarter. We reviewed recent news in novel therapies and implications for the field of rare disease (recent Huntington's Disease Ph2 results for AMT-130), along with new FDA guidance on Rare Disease Drug Dev (<https://www.fda.gov/news-events/press-announcements/fda-advances-rare-disease-drug-development-new-evidence-principles>). Several ideas were discussed for potential programs at 2027 ISCTM conference pertinent to ODWG.