

Resource List for Rare Disease Patient Registries

Kelly Trout, BSN, RN
Director, Research and Medical Advocacy
International WAGR Syndrome Association
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- **How-To Articles, Books, Webinars, Websites, Forums**
- **Registry Platforms**
- **Registry Databases**

Articles

Recommendations for Improving the Quality of Rare Disease Registries

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6121483/>

Rare disease registries: potential applications towards impact on development of new drug treatments

<https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0836-0>

Global Genes RARE Toolkit: Understanding Rare Disease Registries:

<https://globalgenes.org/toolkits/understanding-rare-disease-registries-2/understanding-registries/>

"Golden Age for Rare Diseases" Interview with Marshall Summar MD, April, 2014
Marshall L. Summar, MD, Chief of the Division of Genetics and Metabolism at Children's National Medical Center in Washington, DC

<http://www.medscape.com/viewarticle/823589>

From patients to partners: participant-centric initiatives in biomedical research

Nat Rev Genet. Apr 3, 2012; 13(5): 371–376.

http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3806497/#__ffn_sectitle

How disease advocacy organizations participate in clinical research: a survey of genetic organizations.

Genet Med. 2012 Feb;14(2):223-8. doi: 10.1038/gim.0b013e3182310ba0.

Landy DC1, Brinich MA, Colten ME, Horn EJ, Terry SF, Sharp RR.

<http://www.nature.com/gim/journal/v14/n2/full/gim0b013e3182310ba0a.html>

Applying Standards to Public Health: An Information Model for a Global Rare-Diseases Registry

Robert A. Jenders, MD, MS1, Clement J. McDonald, MD1, Yaffa Rubinstein, PhD, Stephen C. Groft, PharmD National Library of Medicine and Office of Rare Diseases Research, National Institutes of Health, Bethesda, MD

<https://grdr.ncats.nih.gov/files/CDEPosterPresentation.pdf>

Informed consent process for patient participation in rare disease registries linked to biorepositories

<https://grdr.ncats.nih.gov/files/RubinsteinIC2011.pdf>

Informed consent and patient registry for the rare disease community: Editorial

<https://grdr.ncats.nih.gov/files/GradyIC2011.pdf>

The European Cystic Fibrosis Society Patient Registry: valuable lessons learned on how to sustain a disease registry Orphanet J Rare Dis. 2014

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4066270/>

The NIH Office of Rare Diseases Research patient registry Standard: a report from the University of New Mexico's Oculopharyngeal Muscular Dystrophy Patient Registry AMIA Annu Symp Proc. 2013

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3900177/>

The Role of Patient-Powered Registries and Research Networks

Community Forum White Papers Thomas A Workman, PhD. American Institutes for Research Rockville (MD): Agency for Healthcare Research and Quality (US); 2013 Sep. Report No.: AHRQ 13-EHC124-EF

<http://www.ncbi.nlm.nih.gov/books/NBK164513/>

Recommendations for Improving the Quality of Rare Disease Registries

<https://www.mdpi.com/1660-4601/15/8/1644/htm?platform=hootsuite>

Books

Developing a Patient Registry: A Practical Guide

<http://www.mrcg.ie/go/publications/guides/developing-a-patient-registry-a-practical-guide>

"Registries for Evaluating Patient Outcomes: A User's Guide"

Agency for Healthcare Research and Quality (AHRQ)

Extremely comprehensive guide from the Agency for Healthcare Research and Quality. **Free**

<http://effectivehealthcare.ahrq.gov/index.cfm/search-for-guides-reviews-and-reports/?productid=1897&pageaction=displayproduct>

Webinars

Key Considerations for Planning and Building a Robust Patient Registry

RARE Webinar, October 2013

http://globalgenes.wpengine.netdna-cdn.com/wp-content/uploads/UnderstandingRegistriesWebinar_part2_slides.pdf

Global Genes Webinar Series

https://globalgenes.org/rare_webinar_series_understanding_rare_disease_registries/

Platforms

CoRDS: Coordination of Rare Diseases at Sanford

<http://www.sanfordresearch.org/SpecialPrograms/cords/>

NORD: National Organization for Rare Disorders

<https://rarediseases.org/for-patient-organizations/ways-partner/patient-registries/>

Genetic Alliance

<http://www.geneticalliance.org/programs/biotrust/peer>

RedCap

<https://www.project-redcap.org/>

Alta Voice (formerly Patient Crossroads)

<https://www.invitae.com/en/patient-insights-network/>

Websites

NCATS, National Center for Advancing Translational Science, National Institutes of Health “Toolkit for Patient-focused Therapy Development”

Starting a Patient Registry:

<https://rarediseases.info.nih.gov/toolkit/pages/55/discovery>

Genetic Alliance

<http://www.geneticalliance.org/programs/biotrust/nets/content/registries>

RaDar (formerly Global Rare Diseases Registry, GRDR)

<https://ncats.nih.gov/radar>

National Institutes of Health/NCATS

<https://ncats.nih.gov/clinical/registries>

Eurordis

<https://www.eurordis.org/publication/rare-disease-patient-registries>

Ongoing News and Forums

Rare Disease Patient Registry Coordinators (Facebook forum)

<https://www.facebook.com/groups/rarediseasepatientregistrycoordinators/about/>

BioBank Bulletin Archive | GeneticAlliance.org

Sign up here: <http://www.geneticalliance.org>

Registry Databases

List your registry on these to help researchers find you

ROPR: Registry of Patient Registries (US, Agency for Healthcare Research and Quality)

<https://patientregistry.ahrq.gov/>

RD-Connect: EU/Worldwide

<http://rd-connect.eu/>

The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers

<https://www.nature.com/articles/s41431-017-0085-z>