

X-Linked Agammaglobulinemia Marks the Spot: Rare Diseases in Evidence-Based Practice

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1. What are Rare Diseases?

A disease or condition that affects fewer than 200,000 people in the United States is defined as a "rare disease." There are as many as 8,000 identified rare diseases that affect between 25-30 million Americans. In perspective, a single rare disease affects less than 0.06% of the US population, but collectively nearly 10% of the American population is affected by rare diseases.

Rare diseases are difficult to generalize. The majority are life threatening or fatal. More than 80% of rare diseases are genetic or have a genetic component. Some rare diseases only have a single-digit number of reported cases - others have tens of thousands. All of these characteristics make rare diseases challenging to work with.

The 1983 Orphan Drug Act provides incentives for companies to develop drugs for rare diseases, and the FDA has approved over 350 drug uses under this act. The NIH Office of Rare Diseases Research coordinates and supports rare diseases research, responds to research opportunities for rare diseases, and provides information on rare diseases.

4. References

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- National Research Council. *Rare Diseases and Orphan Products: Accelerating Research and Development*. Washington, DC: The National Academies Press, 2011.
- Sackett, D. L., W. M. Rosenberg, J. A. Gray, R. B. Haynes, and W. S. Richardson. "Evidence based medicine: what it is and what it isn't." *BMJ (Clinical research ed.)* 312 (January 1996): 71-72.

2. Rare Diseases in Evidence-Based Practice

Evidence-based practice integrates "individual clinical expertise with the best available external clinical evidence from systematic research" (Sackett 1996). However, evidence may not be available for rare diseases.

Multiple randomized controlled trials are needed to establish consistent results; however even a single clinical trial may be difficult to conduct due to the small population size of some rare diseases. Even when a suitable sample is found, ethical issues arise when no other treatment options are available and the trial is required to have a placebo control.

Due to the difficulty of conducting clinical trials and due to the small population size, most literature on rare diseases are uncontrolled case reports or observational studies. These literature types are more subject to bias and possibly misinformation. In many cases, rare diseases are often treated based on the clinician's expertise and insight, and the patient's treatment preferences, rather than through evidence-based practice.

3. Rare Diseases Resources

NIH Office of Rare Disease Research's Genetic and Rare Diseases Information Center (GARD)

<http://rarediseases.info.nih.gov/gard/>

GARD is an online portal for more than 6,000 rare and genetic diseases, for both health professionals and patients. Linked resources include ClinicalTrials.gov, eMedicine, GeneReviews, GeneTests, Genetics Home Reference, MedlinePlus, Merck Manuals, Online Mendelian Inheritance in Man, Oprhanet, and PubMed. Information specialists are available to assist with specific questions via telephone, mail, and e-mail.

PubMed: Clinical Queries

<http://pubmed.gov/clinical/>

PubMed is a database of over 21 million biomedical literature citations. PubMed Clinical Queries provides specialized searches for clinical studies, systematic reviews, and medical genetics, which will assist in finding evidence-based citations on rare diseases.

5. Further Information

Please visit <http://nnlm.gov/psr/pdf/rd.pdf> for more information on rare disease resources, or contact Lori Tagawa at ltagawa@library.ucla.edu.



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