

Meeting the emerging information needs of precision medicine: enhancing health librarian's knowledge and search skills to support a new clinical service

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Abstract

“Our future is precision medicine.” Becoming a world leader in the research and clinical applications of precision medicine is a key strategic commitment for St Vincent's Health Network, Sydney. Precision medicine is an emerging approach for disease treatment that takes into account individual variability in genes, environment, and lifestyle.

We asked ourselves: “As a library, what role can we play in advancing our organisation's strategic commitment to precision medicine?” This is the story of how we, as a library, responded to this challenge by building our knowledge of information resources available to support precision medicine.

Background

In early 2017 the Clinical Services Strategy of St Vincent's Integrated Healthcare Campus, Darlinghurst, announced “Our future is precision medicine”. This announcement followed the establishment of the Clinical Genomics Unit at St Vincent's Hospital (SVH) in late 2016. The Clinical Genomics Unit uses cutting-edge genomic testing technology to facilitate precision medicine in order to improve patients' clinical outcomes. Library staff decided in early 2017 that we were interested in building our knowledge of precision medicine in order to support the strategic direction of the organisation.

Precision medicine

Precision medicine is a revolutionary new approach to medicine. It takes into account the genetic makeup, as well as environmental and lifestyle factors of an individual, in order to determine the best possible treatment. Genomic medicine is also able to predict a person's risk of acquiring a disease, particularly cancers, and additionally to measure an individual's response to treatment. Finally, knowledge of an individual's genetic makeup can help in prescribing medicines in a more effective way. Genomic medicine is only possible because of the Human Genome Project, which mapped our complete genomic sequence in 2003 and confirms that humans have approximately 20,000 genes.

Important concepts in precision medicine

There are three terms that have been used interchangeably to describe precision medicine. 'Personalised medicine' is a term that was used previously in describing this discipline but is losing favour to precision medicine. The term 'genomic medicine' is sometimes used where the approach to treatment is based largely on genomic analysis and genomic data and doesn't take into account lifestyle and environmental factors. The new unit at SVH is called the Clinical Genomics Unit and their approach seems to be predominantly genomics-based.

The terminology around precision medicine can be somewhat overwhelming. The National Center for Biotechnology Information (NCBI) has a number of online books with useful glossaries including *Toward Precision Medicine* (National Research Council (U.S.). Committee on A Framework for Developing a New Taxonomy of Disease., 2011). Pharmacogenomics is an emerging subspecialty in the precision medicine framework (Auffray et al., 2016). It is the study of how genes affect a person's response to drugs. Pharmacogenomics combines pharmacology (the science of drugs) and genomics (the study of genes and their functions) to develop effective, safe medications, and doses that will be tailored to a person's genetic makeup.

Another important concept integral to precision medicine is 'whole-genome sequencing'. Whole-genome sequencing is the process by which our genomic makeup is analysed. It takes less than a teaspoon of blood or saliva to sequence a person's genome. The National Human Genome Research Institute tracks the cost of genome sequencing. The cost of sequencing has reduced exponentially and is still falling, making whole-genome sequencing an affordable option for testing, treatment, and diagnosis. (Wetterstrand, 2016).

Our journey

Our first steps in learning about precision medicine were via e-learning and reviewing the literature. We undertook a number of literature searches around several topics of interest. Unsurprisingly, and because the discipline is new and emerging, there is not much literature around precision medicine from a librarian perspective. One article of interest was "Hospital Library Support of Precision Medicine" by Helen-Ann Brown Epstein (2016). Brown Epstein provides a useful overview of the genomic databases available on the NCBI platform, as well as some search examples for genomic medicine. She also previously published an article on the topic of library support for bioinformatics (Brown Epstein, 2012). Other articles of interest were on libraries and bioinformatics (Geer & Rein, 2006; Li, Chen, & Clintworth, 2013), the interplay of precision medicine and evidence-based medicine (Beckmann & Lew, 2016; Khoury et al., 2008), and precision medicine and big data (Beckmann & Lew, 2016).

We undertook e-learning, including viewing relevant YouTube videos and MLA webinars. NCBI has a wealth of tutorials for each available database. Some of the databases require biomedical knowledge that is above and beyond our skills. Since precision and genomic medicine is an important emerging area at St Vincent's, there have been many interesting presentations at Grand Rounds and a special lecture series on genomics. We attended as many sessions as we could. Each lecture provided a new perspective and built on our knowledge. We were able to network with the clinicians and scientists from the Clinical Genomics Unit who are at the cutting edge of this new and rapidly expanding field.

Searching for precision medicine

Following on from the article by Brown Epstein (2016), we were interested in searching effectively for topics in precision and genomic medicine. We identified a number of headings and subheadings in the

medical bibliographic databases that are particularly relevant for precision medicine. The MeSH subheading “*genetics*” is probably the most obvious and well-used heading, and can be searched with a disease, condition, or gene of interest. Headings such as *molecular diagnosis* (EmTree) and *molecular diagnostic techniques* (MeSH) are important for cancer or oncology genomics. *Pharmacogenomic testing* (MeSH) and *pharmacogenomics* (EmTree) are useful headings to capture articles on the pharmacogenomic aspects of precision medicine. We have been using the Medical Genetics Filter, which is a series of specialised topic filters within the [PubMed Clinical Queries](#) page. Finally, we have been exploring the Public Health Genomics Knowledge Base ([PHGKB](#)), which includes the HUman GEnome (HuGE) Navigator, a searchable online knowledge base in human genome epidemiology.

Developing a precision medicine library guide

Our aim in developing a precision medicine library guide was twofold. First, developing the guide provided us with a goal of learning about the available resources and how they worked. We also wanted to develop a product that would be useful to our clinicians in years to come.

The development of our guide involved several steps. First, we tried to identify genomic databases and resources available at reputable sites including NCBI, Centers for Disease Control (CDC), and the National Library of Medicine. We were also interested in any available Australian resources and other specialist international sites. Another important step was to locate other genomic or precision medicine library guides. We particularly valued the University of Washington, Health Sciences Library’s *Genetics and Medicine Guide*. It was of interest to see how other institutions categorised or grouped their recommended resources.

At this point, we invited Kathy Wu, Clinical Geneticist, to share her favourite genomic databases, resources, and websites with us. She viewed our pilot guide and critiqued our resources. At the end of this consultation, we were confident that we had included the most useful resources for our clinicians. *Appendix 1* highlights the genomics and precision medicine resources that Kathy Wu and the Clinical Genomics Unit team use on a daily basis.

Implications for libraries

Are we able to imagine any implications with respect to the looming era of precision medicine for libraries, information specialists, and librarians? Allow us to juxtapose the possible impacts of precision medicine with the recent history of evidence-based medicine (EBM). The rise of evidence-based practice and its associated culture of systematic reviews has been important for libraries. Our expert knowledge of searching the bibliographic literature and beyond has been sought after by those conducting Cochrane Reviews and other systematic reviews. We have strategically placed ourselves as an important cog in the evidence machine. How then will the rise of precision medicine affect libraries? Will our workload rise, as it has with EBM, or will our expertise be bypassed because many of the resources are highly technical and require expert biomedical knowledge? Will medical librarians be required to obtain qualifications in bioinformatics? Geer and Rein (2006) discuss the possibilities of collaborative relationships between libraries and bioinformatics centres. Khoury et al. (2008) ask if precision medicine will be required to adhere to the same standards of evidence or whether a new model of evidence will emerge with a better fit for the genomic components of precision medicine.

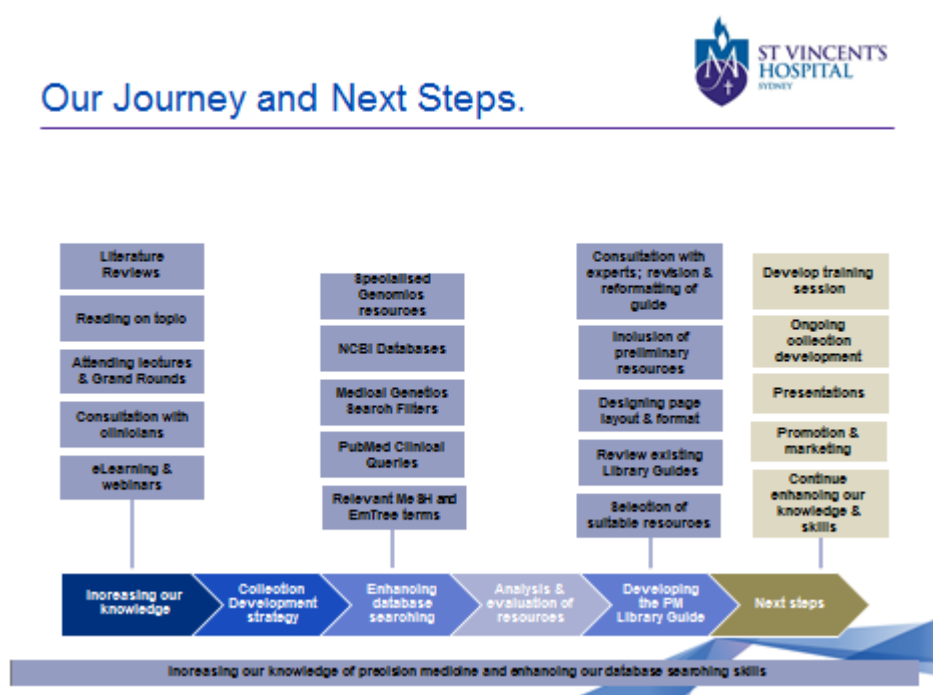
Next steps

Our next steps include a plan to further refine our library guide and develop an orientation and training session for the major resources in the guide. We also plan to deliver further presentations in 2018 including at Grand Rounds, Research Week, and as part of a special lecture series on genomics at St Vincent's. Promoting our library guide, developing our expertise, and bolstering our collection development are also important ongoing commitments. Our journey has only just begun. See *Appendix 2* for a timeline summary of our journey.

Appendix 1. Clinical Genomics Unit Recommended Precision Medicine Resources

- OMIM - <https://www.omim.org/>
- GeneReviews - <https://www.ncbi.nlm.nih.gov/books/NBK1116/>
- Orphanet - <http://www.orpha.net/consor/cgi-bin/index.php>
- Genome - <https://www.ncbi.nlm.nih.gov/genome/>
- Genetics Home Reference - <https://ghr.nlm.nih.gov/>
- PharmGKB - <https://www.pharmgkb.org/>
- Centre for Genetics Education - <http://www.genetics.edu.au/>

Appendix 2. Our journey and next steps timeline



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