Genome Alberta RP3 – BioNet Alberta
Pillar 3 – Tool Development Competition
Health Stakeholder Consultations
Thursday, October 10, 2019

Summary Report

Purpose
BioNet Alberta's mission is to strengthen bioinformatics and computational biology (B/CB) capacity in the province. One mechanism by which this will be accomplished is through a funding competition for the development of B/CB tools and pipelines that address currently unmet needs for the health sector of Alberta. On October 10, 2010, in Red Deer, Alberta, Genome Alberta held a stakeholder consultation meeting with representations from Alberta Public Laboratories (APL) to inform the development of the program Request for Applications (RFA). Specifically, Genome Alberta sought input on the gaps and challenges in clinical 'big data' analyses in the province, what resources exist and are being used, and how the tool development competition BioNet Alberta may improve clinical genomic data analyses. This summary report details the outcomes and highlights from this meeting.

Attendees:
Chair: Ryan Mercer, Research Program Manager, Genome Alberta

Alberta Public Laboratories (APL)

- Graham Tipples, Medical-Scientific Director, Public Health
- Dennis Bulman, Medical-Scientific Director, Genetics & Genomics
- Imran Mirza, Provincial Medical Lead – Molecular Pathology
- Faisal Khan, Associate Clinical Director of Tissue Typing and Molecular Haematology
- Tarah Lynch, Assistant Clinical Professor, Pathology and Laboratory Medicine
- Cheryl Mather, Director of Molecular Pathology
- Sherry Taylor, Molecular Geneticist and North Sector Medical Lead, Genetics & Genomics
- Robert Tomaszewski, Laboratory Scientist (in lieu of Stacey Hume)
- Rehan Faridi, Laboratory Scientist – Molecular Hematology
- Paulo Nuin, Clinical Bioinformatician
- Matthew Croxen, Public Health Genomics Lead

Observers

- Athan Zovoilis, BioNet Alberta Network Lead, University of Lethbridge
- Eric Merzetti, BioNet Alberta Network Manager, University of Lethbridge
- Gijs van Rooijen, Chief Scientific Officer, Genome Alberta
- Matt Bryman, Director of Programs, Genome Alberta
- Niall Kerrigan, Senior Program Officer, Genome Alberta
- Adam Kirkby, Program Officer, Genome Alberta
- Sunil Rajput, Program Manager, Alberta Innovates

B/CB Challenges for APL

The consultation revealed numerous challenges for APL in advancing clinical genomics and the associated bioinformatics in the province. Many of these are system-level challenges for how health care data is captured, analyzed, and results delivered; however, there still remains tangible, incremental improvements that could be realized through the BioNet Alberta funding program that helps move the system towards the future of precision medicine with respect to genomics and bioinformatics. The future goals of healthcare focus on 'Predict and Prevent' as opposed to 'Diagnose and Treat'; genomics and bioinformatics will play a substantial role in providing predictive power.

System Challenges - Infrastructure, Access, and Security

Feedback from this stakeholder group highlighted significant limitations of the Alberta Health Services Information Technology (IT) landscape and digital infrastructure (e.g. firewall). These limitations result in researchers sinking large amounts of time into issues as simple as installing necessary software for completing data analyses. In order for clinical genomics and bioinformatics to be conducted efficiently, there needs to be a paradigm shift in the delivery of IT services for APL towards more advanced and diversified computational software.

The necessity for enhanced data security and encryption adds another layer of complexity. While the management and analyses of clinical genomic and meta-data are understandable subject to stringent privacy policies, improving data transfer and access within and outside of the AHS system is key for developing more robust analytical approaches. The recent implementation of the very unique Connect Care model across the province may alleviate some data access challenges for groups within AHS. However, the inclusion of genomic data in the province-wide system has not been fully conceived.

APL also faces challenges in data storage and computing power. The stakeholder groups acknowledge that with the growing demand for clinical genomics services in the future, there will be a challenge in storing the masses of raw and analyzed data. The anticipated scale-up in the province could see more than 30,000 genomes be sequenced per month, necessitating numerous high-throughput instruments, ample computing resource, and robust bioinformatic services. Suggestions for improvement included the use of off-site, clinically validated servers that will allow rapid turn-around on clinician requested genomic testing. As well, a defined policy around the necessities for data storage and archiving could reduce the size of data repositories while adhering to regulatory guidelines. Comments suggested that holding onto patient data forever may cost more than simply re-sequencing as technology evolves and the cost of genomic testing continues to decrease.

Bottlenecks – People and Clinical Validation

Much like other sectors, a current bottleneck for processing clinical genomic data is in number of qualified (e.g. Ph.D.) personnel to handle the demand. The end-users (e.g. physicians) of the data outputs will never be the technically proficient individuals analyzing and interpreting the data. The complex genomic data needs to be interpreted efficiently and accurately, then provided to the end-users in a form that is easy to understand. Stakeholders suggestions for increasing trained individuals includes supporting fellowships for cross-disciplinary learning; the biologist must understand data science and the data scientist must understand the biological implications of a genome sequence.

Bioinformatics in traditional research environment is quite different than those within a clinical application. Research bioinformatic tools can be newly designed and developed continually, while clinical tools must deliver routine and reliable outputs. Clinical bioinformatic tools need also receive clinical validation and updating creates challenges for ongoing validation and version control. Similarly, sequencing instruments must also be accredited, meaning resources currently available in research labs are not suitable for clinical applications. Access to validated tools and accredited equipment is a current bottleneck for genomics and bioinformatics.

Improving B/CB Approaches

Following stakeholder consultations, it is evident that '*tools*' may be more appropriately described as '*Enablers*' that allow users to overcome limitations in understanding, analyzing and drawing conclusions from collected datasets. This may include coding scripts, algorithms, data pipelines, user interfaces, databases and platforms. Stakeholder suggestions for enablers that could be developed under Pillar 3 of BioNet Alberta are listed below:

- Standardization of analytical protocols for handling complex next-generation sequencing data
    - Clear guidelines and approaches for interpreting data quality and output for end-users; ensuring the use of accurate genetic variant databases for interpretation

- Tools for rapid mining large, complex human genetic databases in combination with provincial medical health records; proof-of-principle application of machine learning

- Development and harmonization of data management protocols, databases, and user-friendly interfaces for disseminating sequencing results

- Automated B/CB workflows to expedite analyses and decrease testing turn-around time; tools for rapid access to stored data and increasing computational efficiency.

- Methods for efficient and effective integration of available patient metadata with genomic and phenotypic information; proof-of-principle application of machine learning

- Improved data sharing standards and pipelines for molecular epidemiology
    - Tools for integrating surveillance data with genomic data to rapidly detect and assess pathogen profiles (e.g. antimicrobial resistance)
    - Robust algorithms for predictive risk modelling
    - Interoperable platforms to facilitate cross-talk between public health groups

- Development of a pre-clinical B/CB testing platform using artificial data and hypothetical case studies – a 'virtual' Alberta Public Lab to assess novel tools, pipelines, workflows

Non-technical enablers:

- Reviewing, establishing, updating policy guidelines pertaining to: data safety and security for genomic testing; harmonized data access, storage and processing; infrastructure improvement; economics for clinical genomics; clinician education; and ethical barrier for academic and clinical genomic research programs

Pillar 3 Program Suggestions

The stakeholders were supportive of academic-clinical partnerships for this program. The collaborative research approach would present some challenges for data access, particularly as metadata holds significant value and context when compared to genomic sequence information. The stakeholder suggested the 18-month project length was too short for clinical implementation, which requires development, validation, support, refinement, additional validation all prior to adoption by APL. This supports the strategy for developing pre-clinical enablers (tools, pipelines, workflows, interfaces, etc.) that would require consideration towards the need for clinical validation. The overall goal would be to tackle small, discernable issues towards impacting the large complex challenging of improve clinical genomics and bioinformatics.