Executive Summary

The Transformation Lab at Intermountain Healthcare in Salt Lake City, Utah, opened in 2013 to create transformative patient care solutions through thoughtful design, development, testing, and validation. Funded by forward-looking strategic collaborators like Intel, the Lab is well positioned to solve quality and care challenges by quickly and effectively bringing innovative technologies to the patient’s bedside.

Intel has a vision to accelerate precision medicine, making it available to everyone. As part of this vision, Intel is working closely with industry and governments worldwide to understand the key challenges that must be overcome to accelerate science, translate results, and deliver them today. Intel is working with the Transformation Lab to understand these challenges and develop tools that integrate genetic data and accelerate its use at the point of care.

One challenge to quality patient care is a series of gaps in the collection, use, and storage of data. Family health histories and other patient-entered data rarely make it into electronic health records (EHRs). Genetic counselors are not integrated with these systems. With family health histories and pedigrees that are scanned from paper forms, complete risk assessments cannot be done. Despite the increased use of clinical genetic testing, most test data is not incorporated into EHRs—at least not in a format usable by computers—and therefore is not available for clinical decision support (CDS). The resulting data silos and incomplete EHRs cause incomplete records for both patients and providers.

This white paper proposes a new patient data-centric model to build applications that:

- **Access** multiple data sources for CDS
- **Are integrated** into the clinical workflow
- **Deliver** meaningful and easy-to-understand information

Current Data Gaps that Prevent Precision Medicine

Precision medicine is the customization of healthcare that accommodates individual differences as far as possible at all stages in the process—from prevention through diagnosis and treatment to post-treatment follow-up.

Patient experiences in the healthcare system usually involve multiple visits with multiple providers at multiple facilities. Even in a highly wired system such as Intermountain Healthcare, gaps exist in the collection and storage of data during this process. It begins with patient-entered data like intake forms and family health histories (data gap 1)
to clinical data collected by one provider but often not available to another. Sadly, EHRs still have not solved the gap between paper-based and electronic-based practices.

The problem only becomes more apparent as we add genetic testing to clinical care practice. With the increased use of genetic testing, genetic counselors are in higher demand. Yet they experience a tremendous data gap. Integrated patient management tools for genetic counselors do not exist (data gap 2). The limited family health histories collected by EHRs do not include a full pedigree and, therefore, do not allow for a complete risk assessment.

Most clinical genetic tests are office send-outs that are not tracked in the EHR or by the lab system (data gap 3). If they are, the results come back as a PDF, not in the form of discrete data for computer processing. When the information gets back to the attending physician, it may not be in a form that is understandable or that has readily apparent, clinically actionable treatments. Also, because genetic test results are stored separately from other data, they are not available for CDS (data gap 4).

Newly discovered knowledge further compounds the situation, making insignificant findings today potentially significant in the future. Clinicians need to be aware of the historical knowledge gaps and be alerted when patient follow-up is needed.

The resulting data silos and incomplete EHRs are holding back the promise of clinical genetics to improve diagnosis and treatment and for precision medicine to become the standard of care.

User-Experience-Driven Design

Intel and the Transformation Lab are working together to further understand these gaps and challenges and to develop tools that integrate genetic data and accelerate its use at the point of care. Figure 1 shows the building blocks of a genomics CDS.

Through a user-experience study (UX), Intel and Intermountain will seek to discover how healthcare professionals make clinical decisions about breast...
cancer, including what information they require and why. The study will also try to understand how healthcare workers assign trust to and validate information. This approach will involve in-depth interviews, shadowing, and think-aloud sessions. An important aspect of this work will be to insert the delivery of meaningful data at the right juncture of the clinical workflow through clinical applications driven by CDS.

**CDS-Driven Clinical Applications**

**Individual-Care Applications**

Individual-care applications are embedded in a care workflow to assist genetic counselors and other clinicians in making decisions. They combine comprehensive, structured data including:

- Family health histories
- Pedigrees
- Imaging
- Screenings
- Past clinical data

When coupled with customized clinical guidelines, this information can form the foundation for a personalized care plan for the patient. It may include use of a risk assessment application to offer a risk profile of the patient for one or more inherited diseases, as well as to recommend genetic testing for the patient or family.

An order entry assistance application can suggest the appropriate tests to determine multiple cancer risks or other genetic diseases. For example, a patient with a positive breast cancer biopsy and BRCA gene mutation is also at risk for ovarian cancer. Knowing whether she carries the BRCA gene mutation may change her ovarian-cancer screening schedule. Such genetic testing can greatly shorten the diagnostic process and help provide timely treatment.

When genetic test results are available, a treatment recommendation application can provide the best course of treatment for the patient, including:

- The most effective drugs
- Treatments to avoid due to a lack of clinical effectiveness based on the patient’s genetic profile

In the case of a positive breast cancer biopsy, if the genetic test is positive for the BRCA gene mutation, the patient may be at risk for a second breast cancer. In such a situation, the patient may choose a mastectomy (removal of breast) instead of a lumpectomy (removal of a discrete lump of breast tissue), thus impacting her breast cancer treatment. The application may also make treatment recommendations for other cancer risks such as removal of an ovarian tumor. For some conditions, it may be advisable to avoid radiation exposure. Thus, the treatment plan should be personalized to the patient.
Data Integration for Precision Medicine

Developing a data architecture begins by collecting and storing both patient and clinical data in a centralized (or linked) repository. Next, combining clinical, family health history, and genetic/genomic data will lead to the creation of new query methodologies, new insights into clinical correlations of these data sources, and the generation of new care guidelines and protocols.

Model Components

Figure 2 shows the components of a clinical workflow model. Using breast cancer as an example, the following components need to be built to integrate genetic data into the clinical workflow:

1. **Notification App** (not on diagram). Upon reaching a certain age, a patient is notified by an EHR-generated message that she should schedule a mammogram.

2. **Patient Data App**. Prior to her mammogram appointment, the patient fills out an electronic intake form for initial screening.

3. **Patient Data App**. Prior to her mammogram appointment, the patient fills out an electronic family health history with a full pedigree for further screening.

4. **Data Store**. Data from apps in numbers 2 and 3 are stored in centralized risk-screening and family health history databases (data gap 1). First data gap is filled, as this data will be available to the new CDS application.

5. **CDS App**. Risk-assessment app schedules high-risk patient visit with genetic counselor.

6. **Clinical App**. Patient goes through further screening with a genetic counselor and, with doctor approval, genetic testing is ordered.

Also, optional family testing is decided (data gap 2). The second data gap is filled, as clinicians and genetic counselors are now integrated into the EHR.

7. **Clinical App**. Genetic/genomic testing is ordered with relevant clinical information for the lab.

8. **Clinical App**. Genetic/genomic testing tracked by EHR. [Data Gap 3] Third data gap is filled as this data will be integrated into the EHR for clinical review by all healthcare providers.

9. **Data Store**. Test result narrative and codified results are stored in centralized database (data gap 4). Fourth data gap is filled, as this data will be available to new CDS application.

10. **CDS App**. New CDS application pulls data from combined clinical, family health history, and genetic/genomic repositories, providing the latest knowledge for results review and recommended treatment.

11. **Knowledge Collection App and Data Store**. Current and newly validated risk algorithms, test ordering guidance, and clinical treatment rules are stored in a knowledge base. Updates to this knowledge feed back into the screening, risk-assessment and CDS applications.

12. **For the Future**. When a whole-genome-sequence data repository comes online, #7, #8 and #10 will become one application.

This model not only fills the gaps and integrates genetic counseling, it also helps find the combination that provides opportunities to:

- **Discover** new connections between the data
- **Develop and validate** new risk algorithms
- **Build** innovative clinical decision support tools

The model can be built as a template for any clinical care process, disease, or medical condition. To achieve the goal of interoperability, standards will be used where available. The solutions in this model should work with any data collection tool, so they can be implemented by other healthcare institutions.

Population-Based Data as a CDS Source

Knowledge can be discovered through powerful, population-based applications by combining clinical, family health history, and risk data. Risk algorithms and models can be developed based on historical case data. And targeted interventions can be made based on the risk profile.

Intermountain data scientists will use the Intel® Analytics Toolkit for Apache Hadoop® software to explore new data relationships in the combined data set to formulate hypothesis and develop new risk algorithms of the combined data sets available at Intermountain. The Intel Analytics Toolkit is a platform that unifies entity based machine learning with an end-to-end graph processing pipeline. It includes powerful algorithms for uncovering relationships hidden in big data and lowers the barrier of entry to at-scale machine learning over graph data (Figure 3).

Large population databases—such as the Utah population database that has pedigree, outcomes, and claims data—can be used in conjunction with clinical data to assess patients who may be at risk. While genetic mutations play a part in disease manifestations, it is also clear that the environment and social factors also contribute. Having the right data in a population health database will enable discovery of these environmental and social factors and the importance they play. Models can be created based on historic treatment and outcomes and then applied in practice to suggest the best course of treatment.
Conclusion

Intermountain’s Transformation Lab and Intel have collaborated to overcome the challenges associated with accessing and utilizing genetic data in healthcare settings, and to develop tools that accelerate its use at the point of care.

The result is a new four-part genetic-data solution:

1. **A centralized** genetic/genomic data repository that is linked to all clinical and patient data
2. **Access** for all clinicians to EHRs, including genetic counselors

3. **Genetic test data** that is incorporated into EHRs in a computer consumable format
4. **Novel and interoperable** CDS tools

This concept leads to new insights into correlations of clinical, family health history, and genetic/genomic data sources. These insights are the generation of new care guidelines and protocols that will improve clinical outcomes. By filling these gaps and better integrating genetic counseling, the combination provides opportunities to:

- **Discover** new connections between the data
- **Develop and validate** new risk algorithms

For more information on healthcare solutions, visit www.intel.com/healthcare or www.intermountainhealthcare.org.
Integrating Genetic Data Into Clinical Workflow with Clinical Decision Support Apps