Transforming personalized medicine through the use of an online risk assessment application and telemedicine: a novel platform for the delivery of genomic healthcare

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for

Family Care Path, Inc.

April 2018

Abstract

Despite the fact that family history is one of the most powerful predictors of someone's risk for disease, this information is rarely collected in a comprehensive manner and even less commonly used to assess an individual's disease risk. Incorporation of a standard process to collect and analyze family health history would directly contribute to better outcomes and lower cost. Family Care Path's mission is to communicate to patients their risk for disease based on family history and ensure access to genetic counseling if indicated.

MyLegacy is a web-based questionnaire asking for personal and family history as well as lifestyle and environmental influences. This application is SMART on FHIR certified and can seamlessly integrate into electronic medical record systems. The information provided by the patient is evaluated via complex algorithms established by the Genomic Medicine Institute at the Cleveland Clinic. Personalized risk levels are calculated for several disorders and provided to patient and physician along with recommendations regarding next steps. Patients at high risk for hereditary disorders are offered genetic counseling through CarePathConnect, our national telegenetic counseling network. Given the small number of genetic counselors in the US, removing the barrier of access to genetic counseling is crucial to ensure that patients receive appropriate genetic testing and adequate information about results and management changes.

Here is how we engage patients directly: We make medicine personal!

Personalized or Precision Medicine is increasingly on the forefront of innovative medicine and there is no question that *this* is the future of healthcare. There is nothing more personal than one's own family and it is well known that family history is one of the most powerful predictors of a person's risk for disease. It is in fact more reliable than many genetic screening tests, blood tests, or imaging. It is however also known that, due to time constraints and a general underappreciation for their full value, family histories are rarely collected in a comprehensive or standardized manner. Furthermore, the information collected is often not adequately analyzed (e.g. via online risk assessment tools) and patients' risks for diseases are therefore frequently not recognized. These are missed opportunities for assessment and proactive disease screening, which can lead to early diagnosis or even prevention of disorders. Disease prevention or detection in an early stage (for example cancer) not only leads to better treatment outcomes, but to overall improved quality of life, prolonged lifespan, decreased disability, and a reduction in lost work days.

At the Genomic Medicine Institute at Cleveland Clinic, it was realized that many patients with personal and/or family histories of cancer or cardiovascular disease (cardiomyopathies, aneurysms) were never seen by a genetics specialists, despite clinical guidelines that recommend genetic evaluation and possibly genetic testing to rule out underlying hereditary conditions. The most common reasons for the missed referrals were: a) time constraints and b) insufficient knowledge regarding which patients to refer to genetics. These conditions prompted the creation of a clinical decision support tool that would analyze patients' information and provide the physician with a risk level for certain conditions, as well as recommendations regarding next steps for the respective patient. This application, called **MyLegacy**, was successfully launched within the Cleveland Clinic, and was subsequently taken outside of the institution by our company, Family Care Path, to make it available to the broader public.

What is MyLegacy?

- MyLegacy is a web-based questionnaire asking for personal and family health history as well as information about environmental and behavioral factors. The questionnaire is completed by the patient, for example before a doctor's visit or as part of a wellness plan.
- The information entered by the patient is analyzed via specific disease algorithms which were established by a team of genetics specialists, oncologists, breast surgeons, cardiac surgeons, and internists at the Cleveland Clinic. The algorithms are based on published guidelines, peer-reviewed articles, and widely used risk assessment tools.
- After the patient has completed the questionnaire, a MyLegacy result report is generated and available to the patient and the referring physician. This result report includes risk levels, explanations of the different conditions, and recommendations for next steps to monitor or assess for the condition of interest. In addition, a family tree (pedigree) outlining the relationships of affected individuals toward each other is outlined for better visualization.
- Risk levels range from population risk (no increased risk compared to the general population) to raised, high/familial, and finally genetic risk. While 'high/familial risk' indicates a possible genetic component within the family that increases the individual's susceptibility for a disease,

'genetic risk' raises concern for a hereditary disorder, in which the risk to be affected may be extremely high (between 25-50% if not yet personally diagnosed with the disease).

- The current version of MyLegacy assesses risk for 12 different conditions, including 7 cancer conditions, 3 cardiovascular disorders, and 2 complex diseases (diabetes and osteoporosis). Additional disease algorithms will be added over time.
- All of the diseases for which risk is assessed in MyLegacy are actionable, which means that early recognition leads to a change in the patient's management so that the disease can either be detected and treated early or maybe even completely prevented.
- Should an individual be at familial/high or even genetic risk for a disorder, a referral to genetic counseling is recommended.
- To provide individuals who have high/familial or genetic risk levels with timely and uncomplicated access to genetic counseling, we founded CarePathConnect, a telegenetic counseling network that is available for genetic counseling, coordination of genetic testing, as well as management adjustment based on genetic testing results.

CarePathConnect – telegenetic counseling network

- The demand for genetic counselors and geneticists in the US currently exceeds supply, and access to genetic counseling can often be a barrier to receiving appropriate genetic testing and counseling. Wait times to be seen by a geneticist or genetic counselor can be extremely long, which is not acceptable for patients who may be at high risk for a hereditary condition.
- We therefore developed **CarePathConnect**, a telegenetic counseling network that provides easy and timely access to genetic counseling for patients with elevated risk levels for hereditary conditions.
- Appointments can be scheduled online and consultations occur via a video medicine platform. Patients have ultimate flexibility and can participate in the genetic counseling session via computer, tablet, or smartphone from the comfort of their home, work, or any other desired location.
- During the initial genetic counseling session, the information provided by the patient in MyLegacy is confirmed and updated as needed. The genetic counselor is then able to determine if genetic testing for a certain condition is indicated.
- The genetic counselor provides all the information needed to help the patient understand if he/she would like to move forward with genetic testing.
- If desired by the patient, our genetic counselors initiate the genetic testing process, assist in obtaining prior authorization, and organize sample acquisition.
- Once genetic testing results are available, the patient is contacted and encouraged to schedule a follow up genetic counseling session to discuss results and possible changes in disease management.

- Finally, a written report summarizing the counseling session, genetic testing results, and management recommendations is generated by the genetic counselor and provided to the physician and referring provider.

What are the requirements for a new application in the healthcare sector from a technical standpoint?

With modern healthcare applications, flexibility is key. Consumers expect to have full access to their personal information on demand. Consider personal banking in the 1980s versus personal banking today. Transactions that could only be completed in person at the bank can now be done remotely on a personal computer or mobile device.

Healthcare is now on a similar trajectory. Consumers expect deeper access to their personal health information when and where they want it. This type of access, through connected health applications, will inevitably lead to higher engagement levels and higher quality health information. Most importantly, consumers will be able to control this information outside the context of any given health system. Their personal health information would follow them from doctor to doctor and be available for them to share with other health apps.

This future is enabled by SMART on FHIR. Applications can manage the patient's identity through single sign on and share their information (with appropriate consent) with other health and wellness partners. In this model, the application can run either as a standalone application or in coordination with an electronic health record. In either case, the application will be based on health messaging standards, and can move seamlessly between environments. At the end of the day, incentives can line up to provide a better user experience, higher quality health information, improved communication (outside of the traditional encounter model), enhanced clinical decision support, and better patient outcomes that can be shown with supporting data.

Summarizing the features that are necessary for the successful implementation of a new application in the healthcare sector:

- Secure and scalable data management
- Standards-based messaging (i.e., FHIR, HL7)
- Integration with multiple electronic health records
- Mobile-first design that adjusts to the user device
- Implementation of modern development techniques and software patterns
- Ease of use for patients and clinicians
- Inline help and user support
- Straightforward reporting

How does our innovative approach change clinical patient care and what are the requirements from a clinical perspective?

In an environment with increasing expectations for physicians to see more and more patient in less time, new technology must add value to patient care and decrease (or at least not increase) clinical workload. From a patient or consumer perspective, at times of sheer endless choices in the app store, a new application needs to offer content that distinguishes it from other, similar products on the market.

Hence, for a new technology to be successful, several key requirements have to be met and our MyLegacy application in conjunction with the telegenetic counseling network CarePathConnect fulfills the following criteria:

There has to be a perceived benefit to both, clinicians as well as patients.

<u>For clinicians:</u>

- Improved quality of care with little or no additional effort since patient-entered data is automatically analyzed and recommendations are provided via the MyLegacy result report (clinical decision support).
- Appointment cancellations and no shows decrease when MyLegacy is completed since patients are more actively involved and there is interest to learn the risk level results.
- Identification of at risk family members may increase patient numbers for physicians.
- Physicians can rely on their patients receiving appropriate genetic testing through CarePathConnect, without having to worry about prior authorization or test result interpretation and management recommendations.
- Increase of downstream revenue for the provider if high-risk patients are adequately identified and appropriately managed.

<u>For patients:</u>

- MyLegacy is well-perceived by patients: they take ownership of their own health and show increased compliance (since they have invested time in questionnaire and family history is very personal).
- Facilitation of access to personalized medicine since management can be tailored to an individual risk profile.
- Easy and timely access to genetic counseling via CarePathConnect if indicated. This surmounts an often-encountered barrier due to limited availability of genetic counselors and long wait times.
- Assistance with choice of correct genetic test, prior authorization, and result interpretation through the genetic counselors at CarePathConnect.
- Better clinical outcomes, quality of life, and increased life span for patient AND family members due to early disease detection and/or disease prevention

The technology needs to be easy to use and flexible.

- MyLegacy is accessible from any device including computer, smartphone, and tablet.
- The questionnaire is clearly structured and technical terms are explained in a comprehensive glossary.
- Data entry process can be interrupted as often as necessary, for example if the patient needs to gather additional information about family history.
- Patient-entered information is saved and can be updated on a yearly basis going forward.

For clinicians, a new application needs to fit into their natural workflows and should not add to their workload.

- The MyLegacy questionnaire will be completed by patients before a visit with their physician.
- While adding a few minutes of time to the initial doctor's visit after MyLegacy has been completed to discuss results, its use has been shown to overall decrease appointment length, while at the same time improving quality of care.
- With seamless integration into the electronic medical record systems, MyLegacy results are easily accessible to the health care provider without adding too many clicks to the process and without having to leave the digital work environment.

The new technology needs to offer the opportunity for research.

- Information entered into MyLegacy and associated risk levels provide valuable clinical data to physicians.
- The combination of genetic testing results with comprehensive phenotypic information creates an easily queried database that can be immensely useful for research and therapy development.

How do we get the broadest use of our new application?

In order to make a new application successful, the technology should be useful in different environments and for broad range of clients. This requires a certain degree of flexibility to customize the application and adjust to different workflows, provider groups, and result recipients. For MyLegacy and CarePathConnect, we target the following different customer groups:

- Hospital systems/health care providers: MyLegacy is used as a clinical decision support tool and helps improve patient care without increasing physician workload. CarePathConnect (telegenetic counseling) is a valuable and necessary addition in places where lack of access to genetic counseling presents a barrier to appropriate care.
- Self-insured employer groups: There is great interest in early detection of disease risk in order to take preventative measures. This saves treatment cost (e.g. no need for

chemotherapy/radiation if cancer is detected at very early stage) and decreases sick days and disability. In addition, through CarePathConnect, inappropriate costly genetic testing is also avoided.

- Wellness programs: Disease prevention is one of the main pillars of wellness programs. Identifying at risk patients and taking appropriate measures (such as diet changes and exercise routines) is facilitated by the MyLegacy application. While nutritionists are often part of the wellness program, genetic counseling is not always easily accessed and this void is filled with CarePathConnect.
- Direct-to-consumer (DTC): This is a rapidly growing market, since general population interest in genetic information is steadily increasing. As opposed to many DTC-offered tests which are not based on sufficient evidence and don't convey clinically actionable information, MyLegacy offers real health information that is used to determine whether genetic testing is really indicated, and which test is most appropriate for the individual. Especially for the DTC population, access to genetic counseling via CarePathConnect is critical to ensure that even outside of a health care environment, appropriate pre- and post-test genetic counseling is provided and informed decisions can be taken.

Conclusion

In summary, the success of an innovative application in the healthcare sector depends on several factors, including novelty, ease of use, added clinical value, compatibility with different electronic devices, integration into electronic health record systems, and broad application possibilities. Family Care Path's risk assessment application MyLegacy, in conjunction with the telegenetic counseling network CarePathConnect, is fulfilling all of these requirements. With ever-increasing numbers of genetic testing, it is almost impossible for patients, physicians, and insurers to decide which genetic test is appropriate, informative, and provides actionable results. Therefore, our foremost goal is to provide a technology that communicates a reliable disease risk assessment to individuals and their health care providers. This in turn allows for appropriate referral to genetic counseling and careful evaluation of the necessity for genetic testing – often a barrier not only for patients but also for insurers, which is easily overcome by easy access to telegenetic counseling through CarePathConnect. Overall, this is a promising and innovative approach to improve access to and quality of personalized medicine on a global scale due to its integrative nature and technological flexibility.