Web Development of Direct-to-consumer Genetics Testing

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***Abstract*—As the cost of genetic testing becomes more affordable each year, direct-to-customer (DTC) genetic testing services witness rapid market growth. This has encouraged the development of an easy-to-use website application to optimize potential customers to obtain informed choice regarding the offered DTC genetic testing as well as purchasing information. We present a wireframing process as a part of the agile software development process to build a web portal prototype for an Indonesia-based genetic testing service called DNAku. The approach in building the prototype used in this case is a part of the Agile Software Development Method. In brief, the wireframing stage resulted in this work consists of three main important outcomes. The first one is the use case diagram as a blueprint of this web-based system. A site map is then proposed as an extended version of the use case diagram which visualizes the whole pages with the links. Finally, based on this site map a series of page mock-ups are designed using Hyper Text Marking Language (HTML) and Cascading Style Sheets (CSS). The result of the prototype can be used as the basis for the next development stage, which is the coding stage based on the collected feedback from users. Considering the continuous application development via the wireframing method by taking into account the cycle of feedbacks from involved parties, this approach can strengthen the infrastructure to sustain DTC genetic business model, which has a niche market in Indonesia.**

Keywords—agile, genetic testing, prototype, website development, wireframing

I. INTRODUCTION

The completion of the human genome project in 2003 has accelerated the promising progress of genomic medicine and research for clinical and public health purposes to plan effective strategies to understand, diagnose, prevent, and treat diseases based on genetic profiles [1]–[3]. The implementation of genomics in public health has been widely practiced in multiple levels such as personal level, families, organizations, communities, and even many countries began to incorporate it as a part of national public health agenda and national research effort [4]–[8]. Furthermore, this also revolutionizes the era of personalized medicine, for example, we can gather personal medical information from genomic profiles offered by genetic testing services that are marketed toward customers without directly involving a health care provider [9]. This is known as direct-to-consumer (DTC) genetic testing and provides customers to access their genomic profile by analyzing single nucleotide polymorphisms (SNPs) that may be related to a particular disease or traits [10]. Apart from obtaining health and disease risk information, the tests are usually taken by customers to know their ancestry information, kinship, and lifestyle factors (nutrition and dietary pattern, fitness, skincare, etc.) [11].

DTC genetic testing can be beneficial to inform customers about their genetic profile with affordable cost than the genetic testing from health care providers [12][13]. Further, personalized testing may help to raise awareness about their health and potential disease risk, including genetic diseases, and promote healthier lifestyle changes based on the testing results. In addition, DTC genetic testing provides customers to manage the privacy of the results, which can be accessed from their account on particular platforms (e.g. official website, email, etc.) [13].

As the costs associated with genetic testing have dropped significantly, the commercial companies offering such services have seen a surge of popularity nowadays, for example, 23andMe, Ancestry.com, Gene by Gene, Living DNA, and more. The expansion of the internet is a medium for which a wide variety of personalized genetic testing products are advertised and sold. To facilitate customers with detailed information about the DTC genetic testing services and allow the registered customers to order and customize a product through the Internet, a web-based portal is a convenient platform that needs to be developed by the company. Also, the website application provides a medium for consulting the test results to customers [14]. Principally, after registering online through the company's website, the

company will send a deoxyribonucleic acid (DNA) test kit to customers for collecting their DNA samples (blood, hair, tissue, saliva, etc.). Next, the customers send the sample to the lab to analyze and then receive a report of the testing result after a period of time.

In this paper, we propose a prototype of the wireframing process to develop a website for DTC genetic testing services, employing a concrete example from the development of DNAku website. DNAku is the latest DTC genetic testing service launched in 2019 by Genetics Indonesia, an Indonesia-based genetic company [15]. Personalized genetic testing is considered a new service in Indonesia. Therefore, interactive features must be integrated into their website development process in such a way that allows the information flow and adapts with changes based on all parties involved in the website development, including feedback from users. The main advantage of the wireframing approach in this case is the ability for continuous development to consider changes based on suggestions from the project team and also potential users.

II. Related Works

The web applications in clinical and genetic studies usually consist of a centralized database and a software application for allowing data management and data analysis that can be accessed and shared by other researchers and clinicians. Such applications have been implemented for various tasks such as genetic studies in agriculture [16]–[18], a platform for medical records management and monitoring in healthcare practice [19]–[23], and also online learning applications for disease detection and health education [24]–[26].

Specifically, the prospects of online genetic testing prompt companies to develop and design a web portal that can support their model business. For example, Gembiosoft company, a Spain-based genetic company, designed a web application called GenesLove.me that provides online capabilities for DTC genetic testing customers according to a study by Roman et al. [27]. The web application is based on the Business Process Modelling (BPMN) and Conceptual Modelling (CM) that aims to evaluate the process and the participants involved and improve the data management for guaranteeing personalized medicine. Besides, many companies design a website that allows users to share and discuss the testing results via an online forum. It is mentioned that public engagement via digital participation with genetic testing promotes self-discovery and even a social belonging for users who share their ancestry testing results [28]. Ancestry.com is one of the largest private genealogy companies that provide ancestry estimation testing. Their official website collects digitized and indexed historical records made available online to infer family histories and family trees [29]. The website contains interactive features that allow users to share the testing results and discuss them in question and answer (Q&A) forum.

III. Research Methodology

Wireframing stage is included in the initial stage in the whole Agile Software Development approach in building a production web-based DTC genetics testing portal [30]. This approach is the most common methodology used in developing software [17], [31]. The main feature of this approach is iterative development which consistently considers all changes based on the communication among all parties in the development process, such as project owner, developer, designer, etc. By continuously adapting to every change proposed by the team, including from the potential users, this approach allows all requirements are considered to not only make the software functionally established, but also easily used by the users. This approach is the opposite of the traditional waterfall software development life cycle which harder to accommodate the changes during the process [32].

More specifically, SCRUM as the most popular agile software development framework was applied in this software development process [17], [31], [33], [34]. SCRUM can help the team to deliver the development progress gradually by adapting to every change. As the result, several versions of the software, including the prototype from wireframing phase, can be developed which represent the iterative process within this framework. The whole SCRUM diagram can be seen in Fig 1.

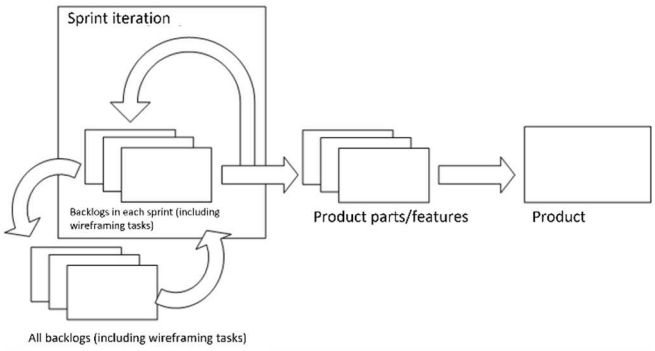

Fig. 1. SCRUM Diagram.

As a best practice in an agile software development pipeline, wireframing is one crucial stage at the beginning of the journey after the requirements have been captured [35]. Wireframing is also part of the design thinking process as the reference for developers in building applications from a design point of view [31], [36]. One main reason why this stage becomes crucial is to make sure the application can meet user expectations from the beginning in terms of information flow, user interface, and design, before beginning the costly and time expensive coding stage. In this stage, a use case diagram as a summarization of the user requirements is built as the basis of the wireframing process. This use case diagram then can be expanded to a site map diagram that can visualize all pages needed to support all items in the use case diagram. This diagram can also be used as a general preview of the whole user journey. Finally, a series of design mock-ups based on the proposed site map can be developed as a prototype of each page. From that we started building a detailed wireframe of every individual page.

The wireframing process in an individual page needs to convey hierarchical structure to achieve the goal of optimizing users' experience when they navigate the page. Next, the final design was implemented using Hyper Text Marking Language (HTML) to define the structure of web page. The design presentation of each page was performed using Cascading Style Sheets (CSS) to set the layout, colors, and fonts of text. The main focus of this prototype is to show the users the journey they can have when using this application. Once the users are satisfied with the flow of the system then the developing process can be continued to the coding stage.

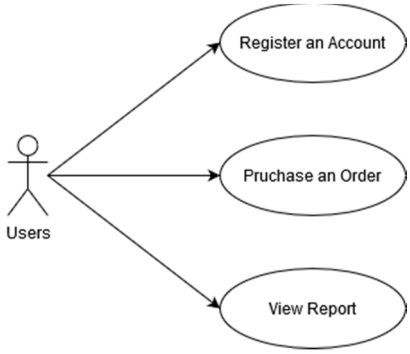

Fig. 2. User case diagram

IV. RESULT AND DISCUSSION

There were three main functionalities focused on the development of this prototype. The first one was the user registration section. The users can use this section to register their accounts to begin the purchasing process. Once the users successfully register their accounts, then they can start to purchase the genetics test services provided on the web portal. The last part was the section for the users to review their genetic testing report. Fig. 2 depicts the use case diagram for these three functionalities.

Based on this use case diagram, a more detailed site map was then proposed to represent each page that needs to be built. Fig. 3 shows the whole site map consists of 10 pages. This site map was the guideline for the web design team to build mock-ups of the system to show the user interface for each page. There were several constraints in designing this web portal based on user requirements. The first thing is given the main target market for this service is Indonesian people, then the language used in this web portal is in Bahasa Indonesia. The other reason was that genetics testing is considered a new thing for Indonesian people, so it is important to deliver the information in their native language with a casual style to be easily understood. However, in this paper, we present the translated version of the wireframing results.

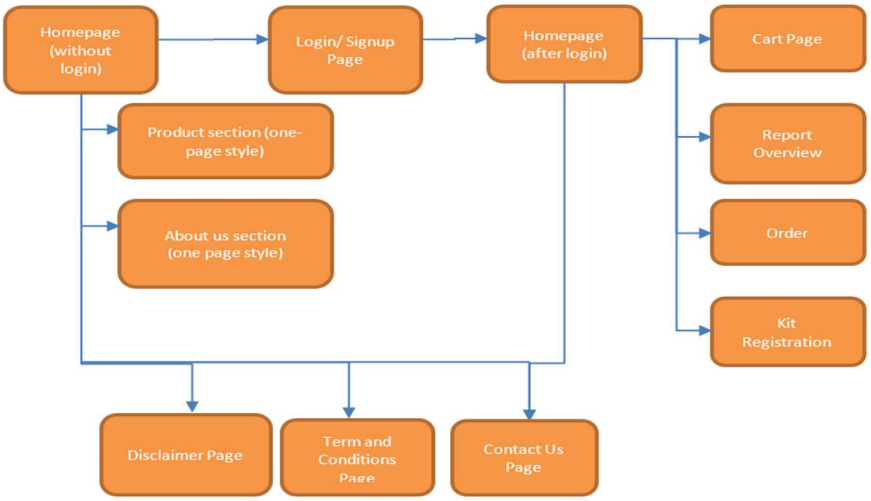

Fig. 3. Proposed Site Maps.

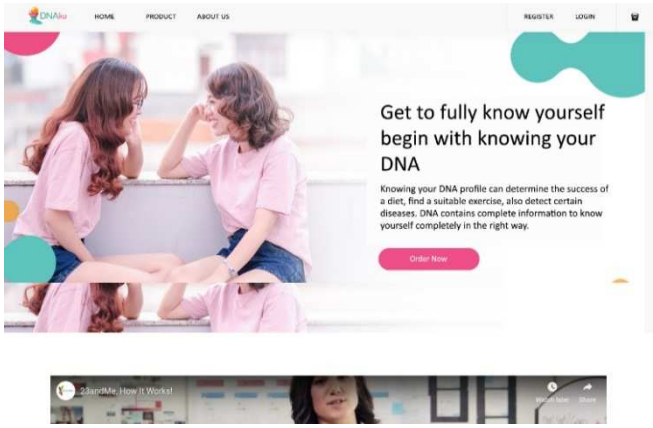

Fig. 4. First homepage (before a login process)

Potential customers can be accessed the website application using a link address of https://dnaku.id/. There are two different homepages in this proposed sitemap based on the authentication status. The homepage for an unauthorized user (i.e. user without a registered account) contains only product description and “about us” sections in a one-page style as can be seen in Fig. 4. The users must register their accounts to purchase genetic testing services. After clicking the order button, the new users are directed to the registration page (shown in Fig. 5) and require completing the necessary information before choosing and purchasing an order.

Moreover, user can make two kinds of purchases according to the business aspect’s requirements. The first one is self-purchase, and the second one is a gift purchase that can be intended for other people such as relatives or friends. As a

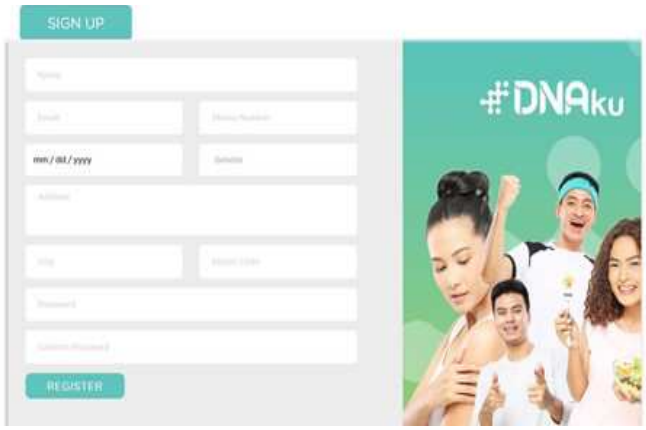

Fig. 5. Account registration page

consequence, on the cart page, as shown in Fig. 6, the users can specify their preferences by selecting the appropriate selection. Additionally, for a gift purchase, an intended name has to be filled to the form as the sample data. After a successful order, the users can track the status of the report by going to the report page. Fig. 7 illustrates the interface of this page which consists of a panel to list all available products and another panel to place the detailed information of the report. This includes the stages of the genetic data processing carried out by DNAku and the availability of the report if the testing is finished. If the report is not available yet, then there is information shows up to notify the users. Once the report is available, the page also provides a feature to save the report as a Portable Document Format (PDF) file by the users to be used for their desire and a documentation for consultation of the result with medical experts.

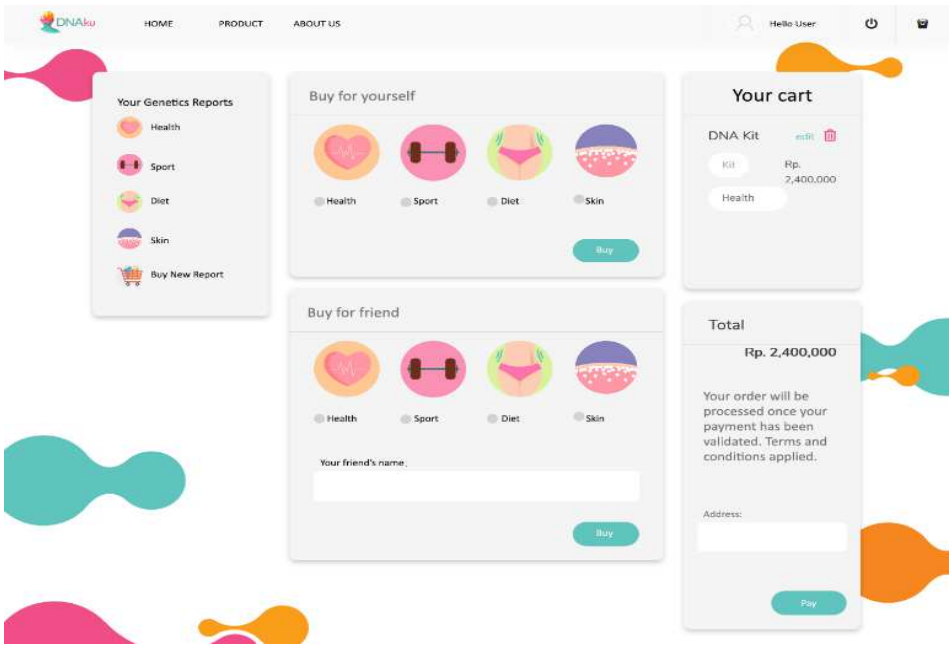

Fig. 6. Cart page

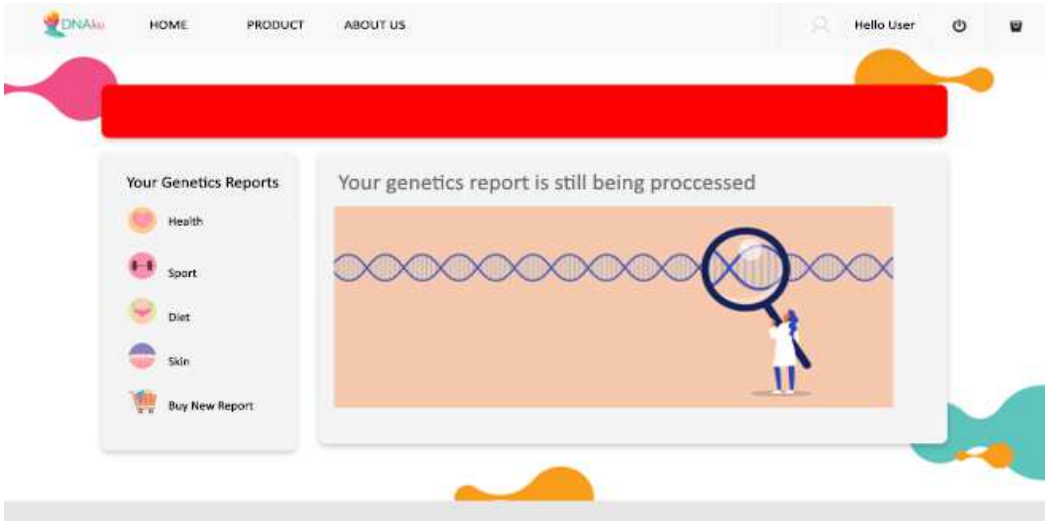

Fig. 7. Report page

After finishing the purchase stage, the user will be directed to a suggestion page in which the users can write feedback and rating in each purchasing stage to improve their navigating experience. The page also provides an option for users who want to postpone giving feedback therefore the system will send a link to the feedback page to users' registered email account so that they can write the feedback anytime. The questions range from the functionality of content and visual/layout design. Since the Indonesian market is mostly not familiar with web-based genetic testing services, facilitating feedback from potential users plays a critical part in iterating the wireframe developments in our website prototype. Moreover, this is also to achieve the user-focused concept in developing the web-based product. This approach also provides a convenient platform for the project team to collaborate on the project vision goal and business needs.

V. CONCLUSION

This paper presents a wireframing result, as a part of the agile software development process of building a DTC genetics testing web portal. This web portal is a vital component to support the business model of this product. The all-online transactions, from purchasing to accessing the report, are the key feature of this brand-new testing which differed this product from other competitors' products in Indonesia. As the central place for interaction between the company and the users, this web portal needs to be built as intuitively as possible. To avoid any mistakes and unnecessary changes, which cost time and money, then the wireframing has been done as a guideline for the upcoming stages. This wireframing task resulted in three outcomes as the translation of gathered users' requirements. These three outcomes were the use case diagram that represents key functionalities needed in this web portal, site maps as the expanded version of the use case diagram, and a series of mock-up pages as the final prototype. Eventually, this prototyping approach can utilize flexible iterative changes from feedback and insights gathered from the stakeholders and even users before starting the coding stage.

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