Applying User Centred Design to Improve the Design of Genomic User Interfaces

Alberto García S.[1][a], Carlos Iñiguez-Jarrín[2][b], Oscar Pastor Lopez[1][c], Daniel Gonzalez-Ibea[3], Estela Pérez-Román[3], Carles Borredà[3], Javier Terol[3], Victoria Ibanez[3] and Manuel Talón[3]

[1]*PROS Research Center, Universitat Politècnica de València, València, Spain*

[2]*Escuela Politécnica Nacional, Quito, Ecuador*

[3]*Centro de Genómica, Instituto Valenciano de Investigaciones Agrarias (IVIA), Moncada, Valencia, Spain*

Keywords: Genomics, User-centred Design, User Interface, GenomIUm.

Abstract: The genomic domain is a complex data environment that has grown exponentially. Several tools have been developed to extract knowledge from this immense amount of data. Knowledge extraction processes depend to a large extent on how easy and intuitive are the user interfaces of the tools that are used by bioinformaticians. However, genomic tools have frequently ignored the design process of their User Interfaces. Consequently, they have important usability problems that complicates knowledge extraction. User Centered Design (UCD) is a design approach that can be used to improve the usability of genomic tools. It consists on putting the user and its real needs at the center of the design process. Improving the usability of these tools will facilitate knowledge extraction. This paper reports the application of the UCD approach to design a tool that improves knowledge extraction processes in a real world-use case. From a general perspective, UCD consists of "user research" and "design solutions". The first one was carried out by conducting UCD techniques, including user interviews and task analysis. The second one was carried out by applying GenomIUm, a pattern-based method that guides the design process of genomic user interfaces. As a fundamental part in the UCD approach, the generated user interfaces were validated by expert bioinformaticians who reported that the complexity of extracting knowledge from genomic data was reduced. We conclude that UCD techniques together with GenomIUm can be a useful strategy to design more usable user interfaces in the genomic domain.

1 INTRODUCTION

Amongst one of the biggest challenges of the century is to get a deep understanding of genomics (Stephens et al., 2015). A so complex domain, containing hundreds of dynamic variables, requires immense efforts to study it. The amount of genomic data that is publicly available has increased considerably over the last decades (Galperin, 2008). This is mainly explained by the reduction in the costs of sequencing genome data (Mardis, 2011) and the increase in the speed of sequencing thanks to Next Generation Sequencing (NGS) technologies (Goodwin et al., 2016). However, being able to generate such amount of data has originated a series of issues that require special attention. The result of these issues is that extracting knowledge in the genomic domain is complex, tedious, slow, and prone to error.

User-centred design (UCD) can be a convenient solution to improve knowledge extraction in the genomic domain. UCD is a product design approach that grounds its design process in information about who will use the product. It is widely recognized that bioinformatics resources suffer from important usability problems (Javahery et al., 2004). Applying UCD can significantly improve the usability of bioinformatics tools, making knowledge extraction more efficient and effective. Although successfully applied in other domains, UCD has been little used in the genomic domain because of its specific particularities. Applying UCD in this domain is complex and requires to overcome a number of additional challenges.

This paper describes our experience applying UCD techniques to develop a bioinformatics tool that

[a] https://orcid.org/0000-0001-5910-4363
[b] https://orcid.org/0000-0003-1338-7542
[c] https://orcid.org/0000-0002-1320-8471

S., A., Iñiguez-Jarrín, C., Lopez, O., Gonzalez-Ibea, D., Pérez-Román, E., Borredà, C., Terol, J., Ibanez, V. and Talón, M.
Applying User Centred Design to Improve the Design of Genomic User Interfaces.
DOI: 10.5220/0010187800250035
In *Proceedings of the 16th International Conference on Evaluation of Novel Approaches to Software Engineering (ENASE 2021)*, pages 25-35
ISBN: 978-989-758-508-1

improves domain knowledge extraction processes. The tool has been developed in a real world-use case along with the collaboration of the *Instituto Valenciano de Investigaciones Agrarias (IVIA)* (Wu et al., 2014; Wu et al., 2018), an agri-food research institute whose work focuses on improving the productivity and sustainability of the citrus agricultural activity. We focused on identifying their key tasks and how to improve them by defining the most convenient User Interfaces (UIs) in terms of usability. To do so, the tool has been developed using GenomIUm (Iñiguez-Jarrin, 2019), a UCD-based framework that provides i) a method to design and implement big data UIs and ii) a catalog of User Interface design patterns to support the process.

To illustrate our work, the paper is structured as follows: Section 2 discusses the state of the art regarding the use of UCD in the genomic domain. Section 3 studies the use case to be improved by identifying the problems to be addressed: the lack of automating and how to visualize the data. Section 4 presents how these problems are addressed by applying GenomIUm to generate the needed bioinformatics tool. Section 5 validates our proposed solution with an evaluation based on user observation. Firstly, we observed how IVIA domain experts interact with the proposed solution to solve a specific genomic analysis exercise they are familiar with. Secondly, we interviewed them to gather valuable feedback. Lastly, Section 6 discusses conclusions and proposes future work.

2 STATE OF THE ART

The ultimate goal of UI design is to produce *usable* UIs that are easy to use and learn and allow users to efficiently perform their tasks and achieve their goals (Rimmer, 2004). However, in complex domains such as bioinformatics, this goal is poorly addressed and there is a growing concern that current approaches are inadequate for this kind of domains (Chilana et al., 2010). Javahery et al. highlight that the complexity of the UIs of bioinformatics resources is higher when they are compared to the interfaces of web sites that people use daily (Javahery et al., 2004). In line with that, Carpenter et. al. suggest that usability should be a more highly valued goal to increase the adoption of bioinformatics tools (Carpenter et al., 2012).

Understanding how users work becomes vital to provide useful UIs in such a complex domain. Understanding what tasks they perform and what workflows they follow allows to better adapt the tools to their specific needs. Svanæs et. al. state that genomics tool UIs should take into account not only the user's needs but also its particular context as a manner of providing more usable solutions (Svanæs et al., 2008). Stevens et. al. conducted a set of surveys of bioinformatics tasks resulting in a task classification to assess the quality of query systems (Stevens et al., 2001). Tran et. al. performed a cross-sectional study of bioinformatics tasks that were documented and proposed as potentially desirable system features in bioinformatics tools (Tran et al., 2004). Rutherford et. al. examined how large DNA sequences are examined and navigated by users to improve the usability of DNA-sequencing navigating tools (Rutherford et al., 2010). All these works provided a better understanding of the unmet needs of genomic domain users.

There has been an explosion in the number of available bioinformatics tools in the last decades. A good example is OMICtools, that provides a catalog of more than 20.000 web-accessible bioinformatics tools (Clément et al., 2018). A common point found among those tools is that their developers tend not to focus on their interfaces or usability aspects (Pavelin et al., 2012). The designed UIs do not consider user's perspective and requirements as the starting point of the design process (Al-Ageel et al., 2015). Consequently, most of the users of these tools find difficult to access the information and too frequently they struggle to find valuable information for their research. They accept tools with poor usability because they use them freely, though these tools do not always provide what they need (Pavelin et al., 2012). A usability testing of bioinformatics tools conducted by Bolchini et al. (Bolchini et al., 2009) reported that usability issues affect the efficiency and effectiveness of bioinformatics work. Several reasons seem to be the underlying cause of not focusing on bioinformatics tool UIs and their usability aspects (Pavelin et al., 2012; de Matos et al., 2013; Chilana et al., 2010):

- Bioinformatics has historically relied on command-line tools and using UCD requires a "cultural shift".
- Bioinformatics data that have to be presented are complex and highly interconnected. Additional technical and scalability constraints have to be considered. Besides, it is a constantly evolving subject whose rules usually have plenty of exceptions.
- Using UCD techniques generates an initial delay in the design process and measuring the impact of applying these techniques is too difficult. UCD techniques improve scientific discovery processes, but "discovery" is an intangible metric and therefore difficult to measure.

- The prior knowledge that is needed to adequately carry out UCD techniques in this domain (human-computer interaction, bioinformatics and computing) creates a gap between domain users and developers.
- The usability validation, crucial to provide successful solutions (Jaspers, 2009), needs to be carried out by skilled UI designers, which is not always possible.

These reasons make the design process of user-friendly UI difficult. Apart from that, authors tend to give more importance to the novelty of the developed tool lessening down usability and UI aspects. Indeed, it is the novelty of the tool its most valued aspect rather than its associated UCD work(Pavelin et al., 2012). In summary, usability and UI aspects have been frequently ignored historically.

However, more recent bioinformatics projects are considering UCD when designing and developing their UIs. A scenario-based visualisation tool to support epidemiological research called ADVISES was developed using UCD methods (Sutcliffe et al., 2010). They used prototyping and storyboarding techniques to analyze user tasks and their domain mental model. The EB-eye search service was redesigned following UCD principles (Valentin et al., 2010). Several user interviews were conducted to gather the initial information and requirements before developing the search service. After developing it, one-to-one usability testing sessions were performed to collect user feedback. The Enzyme Portal was developed after performing a series of user workshops and interviews to identify user needs (de Matos et al., 2013). Afterwards, they tested multiple prototypes until finding an optimal design in terms of navigation and functionality.

In conclusion, having the user as the primary source of information of the UI design and development processes results in multiple benefits (Pavelin et al., 2012). The users will be more likely to use a tool if they guide the design process; having greater access to the data will increase users scientific discoveries. Overall, UCD helps to develop high-quality bioinformatics resources that ease users work and better adapt to their specific needs.

3 PROBLEM STATEMENT

The reported use case consists of applying UCD techniques to develop a bioinformatics tool to aid performing a specific analysis process in the field of genomic citrus plant (variety) improvement. This analysis consists of establishing genotype-phenotype relationships, that is to say, the observable traits in the varieties (phenotype) that are caused by the genetic code (genotype). For instance, the variations in the genetic code that make a variety to be drought resistant. Consequently, it is crucial to properly prioritize (i.e. identify and select) those variations that have an impact in the phenotype. We focus on the prioritization of genetic variations that might have a notorious impact on plant phenotypes. This analysis is a problematic and inefficient process that involves several manual tasks that are difficult, slow to perform, and prone to human failures. These tasks can be grouped into:

Task 1: Select Variety Groups. There are tens of sequenced citrus varieties and it is difficult to work with multiple of them because of the huge amount of data contained on each of them. In order to work with the varieties, bioinformaticians have to select and group them based on specific phenotypes. Two groups are created, one containing varieties that highly express a phenotype of interest and the other one containing varieties that do not express it. For instance, a phenotype of interest is the sweetness of the fruits that a set of sequenced varieties produce.

Task 2: Compare Groups. There are a plethora of variables to consider when filtering the data. Domain experts have to reduce the amount of genomic data by applying several conditions as a previous step before comparing the variety groups. For instance, establishing a quality data threshold or selecting a specific genomic region. They also need a report of the applied filters to manage them easily. Considering the filter conditions, the variety groups have to be compared to extract their differences at a genotype level, i.e. genetic variations. Although applying a single filter or performing simple set operations (e.g. data intersection or subtraction) are challenging but feasible tasks, chaining multiple filters or performing more complex operations are not possible. As the number of varieties involved increases, the complexity and cost of the data filtering task increase dramatically.

Task 3: Visualize. The amount of data obtained after performing Task 2 can become unmanageable and the bioinformaticians require to fluidly examine them to identify potential genetic variations of interest. By "examine" we mean to i) show how the data are distributed based on specific criteria and ii) interact with the data by showing or hiding data columns and performing data.

The generated knowledge is *highly valuable* because it allows modifying citrus varieties so that they can potentially increase or decrease the level of expression of phenotypes of interest. However, as a

consequence of the complexity of the prioritization of genetic variations process reported above, extracting knowledge is complex and requires a considerable effort. The UI design process focused on automating the process and decreasing its complexity so that bioinformaticians can more easily extract knowledge. To accomplish this goal, the three main identified tasks become an entry-point to apply our UCD-oriented solution.

4 PROPOSED SOLUTION

Our proposed solution consists of developing a bioinformatics tool, whose UIs have been designed following a UCD approach. UCD puts the user in the center of the design process to ensure that the resulting UI meets their real needs and interactions. From a general perspective, UCD can be summarized into two main activities: user research and solution production. In the first activity, we have researched our domain expert users by applying UCD techniques such as user observation and task analysis. Observing them while performing the prioritization of genetic variations process is a crucial activity to identify problems related to the data manipulation, detail the high-level tasks identified in the problem statement and determine which UIs should be designed. In the second activity, we have designed the UIs by using the UI design patterns that better address the data manipulation-related problems identified in the previous activity.

4.1 User Research

Domain users have been characterized through several interviews and observing how they work. Identifying and analyzing the tasks involved in the prioritization of genetic variations allowed us to understand both the user mental model (i.e. how they think the variation prioritization process works) and the domain under study. The gathered information has been consolidated in a task model of the envisioned system defined by using Concur Task Trees (CTT) notation (Paternò, 2003) as shown in Figure 1. CTT notation allows to represent the tasks with a chronological and hierarchical structure. Figure 1.1 is the main CTT whilst the "Define filters" and "Examine variation distribution" tasks are detailed in Figures 1.2 and 1.3 respectively for reasons of space. The task model contains the three high-level tasks defined in Section 3 (i.e. select variety groups, compare groups and visualize) decomposed in lower-level tasks.

The **first task**, select variety groups task, consists of defining the two groups of varieties to be compared. Each citrus variety has a set of genetic variations from which some of them are unique and some are shared with other varieties. The system shows the list of available sequenced citrus varieties. Then, the user selects the varieties of interest and adds them to the groups.

The **second task**, compare groups task, consists of two lower-level tasks: define filters and perform comparison. In the first one, the conditions to filter the genetic variations are defined (Figure 1.2). Up to eight filters can be defined from which six are mandatory:

1. Variation type (mandatory, unique): Two types of genetic variations can be compared, namely, Single Nucleotide Polymorphism (SNP) and insertion/deletion (indel). On the one hand, SNPs are changes in the genetic code that only affect one nitrogenous base (A, C, G or T). For example, a variation that changes a C for a T at a given position. On the other hand, indels are genetic variations where the length of the genetic code is altered, either by addition, deletion or both.
2. Set flexibility criteria (mandatory, unique): This filter refers to how restrictive is to accept a variation based on its frequency of appearance among the varieties of a group. By default, only genetic variations that appear in every variety of a group are accepted. However, in some cases this might be too restrictive. The "flexibility" has to do with the ability to filter genetic variations that exist in a subset of the varieties of a group. Such subset is defined by indicating a minimum and maximum threshold of varieties to be considered. There are multiple reasons to do that: working with large groups of varieties, genetic variations wrongly identified in the sequencing process, varieties exhibiting a common phenotype caused by different genetic variations, etc.
3. Quality (mandatory, unique): Because of technological limitations in the sequencing process, genetic variations are complemented with a set of quality indicators that show how reliable they are. This filter allows specifying the quality threshold to accept genetic variations.
4. Annotation impact (mandatory, multiple): Genetic variations are annotated with software to predict their effect and impact at a genomic level (Cingolani et al., 2012). This filter allows specifying the impact and effect under which a variation is accepted. Genetic variations are classified by how significant they are. A variation will be much more relevant if it is predicted to alter a protein's

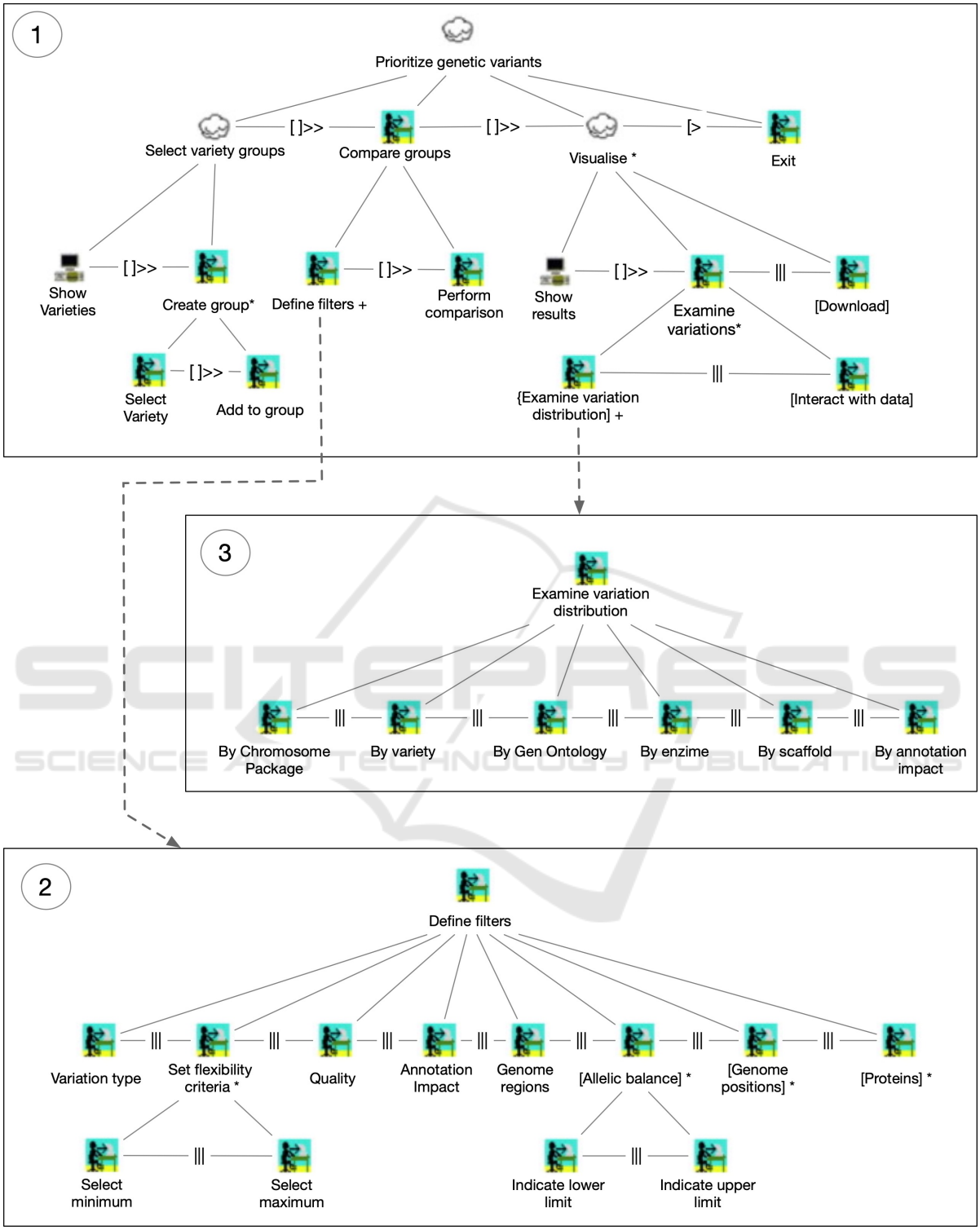

Figure 1: Task analysis.

functionality in a disruptive way (high impact).

5. Genome regions (mandatory): Genetic variations can be located in specific types of regions (intergenic regions, genes, exons, introns, etc) with unique functionality. This filter allows specifying the genomic regions where genetic variations have

to be located to be accepted.

6. Allelic balance (optional, multiple): The analyzed citrus plants are diploid (i.e. their cells has paired chromosomes[1]) so they have two copies of the DNA sequence. When a variation is identified, it can appear in one of these copies or in both. The allele is the sequence, in one of the copies, in the specific position where the variation has been identified. The allelic balance is defined as the ratio of appearance of possible alleles of a variation in the copies of the DNA sequence of a citrus plant. This ratio can range from zero to one. This filter allows specifying multiple lower and upper limit pair values of allelic balance. Only those genetic variations with an allele balance value inside one of the defined ranges will be accepted.
7. Genome positions (optional, multiple): Genetic variations are located in specific positions of the DNA sequence. This filter allows specifying the genomic positions where genetic variations have to be located to be accepted.
8. Proteins (optional, multiple): Some genetic variations affect protein aspects, such as protein's structure or how they work. This filter allows filtering genetic variations based on how they affect a specific protein aspect.

In the second one, perform comparison, the two groups of varieties are compared considering the applied filters. This comparison consists of four operations that the system performs internally: Firstly, those genetic variations that do not pass the defined filters are removed. Secondly, the genetic variations of the first group of varieties are intersected. Thirdly, the genetic variations of the second group of varieties are intersected. Fourthly, the symmetric difference of the genetic variations of the two groups is obtained.

The **third task**, visualize task, consists of examining the data. It involves to i) examine how genetic variations are distributed over multiple criteria, also called passive analysis and ii) interact directly with the data (active analysis). Passive analysis allows users to get a general vision of the data at a glance. To do that, six different visualizations are used:

- By Chromosome package: a visual representation of the genetic variations with their physical location at a chromosome level.
- By variety: number of genetic variations for each variety in the defined groups.
- By Gene Ontology: number of genetic variations for each gene ontology type.

[1]https://www.genome.gov/genetics-glossary/Diploid

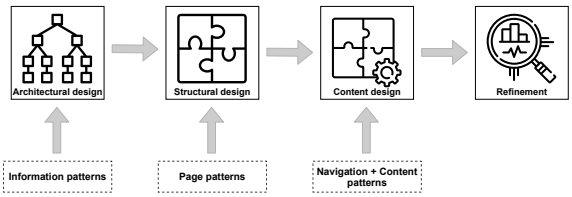

Figure 2: GenomIUm phases.

- By enzyme type: number of genetic variations for each enzyme type according to the type of reaction they catalyze.
- By scaffold: number of genetic variations for each scaffold.
- By annotated impact: number of genetic variations for each annotated impact.

Active analysis allows users to interact with the data in a more complex way. Multiple actions can be performed, combined and chained, including: filtering, grouping and aggregating data, showing and hiding data attributes and performing pivoting operations. Interaction with data can be performed by filtering, grouping and aggregating operations that can be combined and chained.

The characterization of these tasks becomes a foundation that guides the design decisions to generate the UIs that will improve and facilitate the genetic analysis process.

4.2 User Interface Design

So far, we have identified the tasks involved in genomic analysis. Now, our attention focuses on translating those tasks into a tangible UI design. To do that, we focus on developing a key artifact of the design activity: the *conceptual design* (CD) of the UI, which captures the structure and flow of the UI. To define the CD, we have applied a method called *GenomIUm* that has been developed in previous work (Iñiguez-Jarrin, 2019) (see Fig. 2). It is based on Pattern Oriented Design (POD) approach (Javahery and Seffah, 2002) and aims to assist designers in creating the CD of genomics UIs.

GenomIUm takes advantage of the two main characteristics of POD by providing i) a systematic design process and ii) a catalog of interconnected patterns that support the systematic process.

The systematic design process consists of four steps:

1. Architectural Design: This step consists on defining the UIs that will make up the system and their navigation flow. This step is supported by information architecture patterns, which describe system-wide solutions that organize the content to

be displayed by defining high-level presentation units and how they are linked.

2. Structural Design: This steps focuses on establishing the internal structure of each of the UIs defined in the previous step. This step is supported by page patterns, which describe the internal structure (i.e. sectors) of presentation units.
3. Content Design: This step consists of selecting the specific content elements that conform the internal structure of each UI defined in the previous step. This step is supported by navigation and content patterns, which describe the content elements that compose sectors. Each pattern allows users to perform a specific identified task.
4. Refinement: Each design pattern provides a general UI design solution. This step consists of adapting such a general design solution by indicating the visual details of the selected patterns in the previous step according to the specific particularities of the data that is involved in the genomic analysis.

The process is iterative in nature and designers can repeat the steps several times until the CD meets the user needs.

The catalog is structured in several pattern categories, one for each step of the design process and it covers general design problems (i.e. navigation or interface distribution) as well as specific design problems (i.e. visualizing the complete set of chromosomes of a species). Designers can exploit the pattern relationships to create complete or partial UI designs.

The process and its pattern catalog cover the design of the UIs of a complete genomic application. In the following paragraphs, we describe the CD resulting from applying GenomiUm in a joint work with bioinformaticians. Figure 3 shows the designed UI after performing the GenomIUm method.

In Step 1, *Architectural design*, three UIs have been defined based on the tasks analysis (Fig. 1): *Variety Selection UI* for the "Select variety groups" task, *Filter UI* for the "Apply Filters" task and *Visualization UI* for the "Visualize" task. Bioinformaticians performed several UCD activities to guide the definition of the UIs. As an example, figure 4 shows them performing a card sorting session. The defined UIs are connected through the "Sequential" pattern (the UI with the "H" letter indicates the initial UI). This pattern is used when a complex task can be divided into more simple tasks that are performed in a sequential order. It guides bioinformaticians through the three UIs to carry out the "prioritize genetic variations" process.

In Step 2, Structural design, the sectors of the UIs

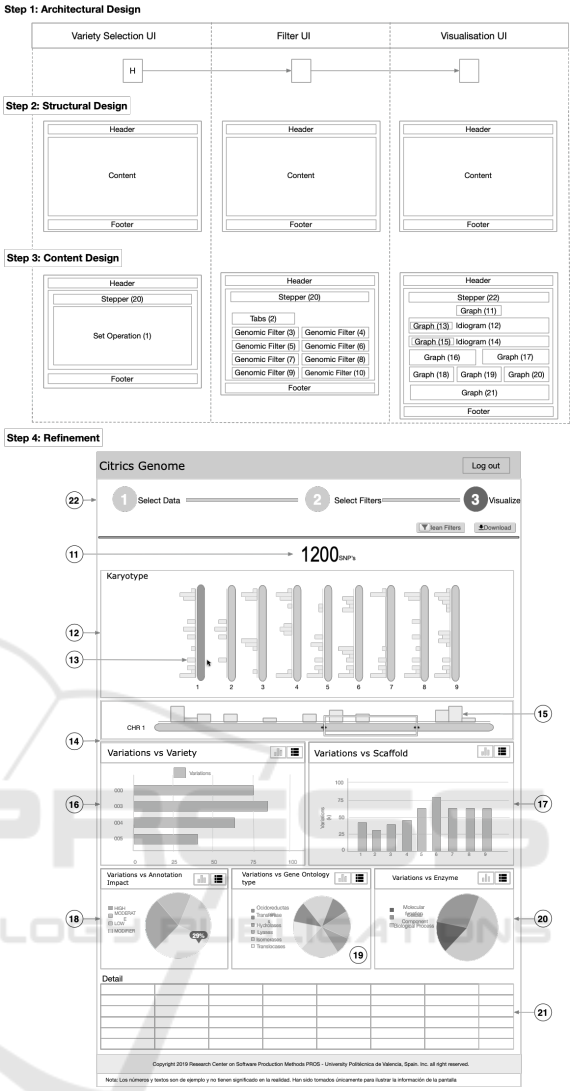

Figure 3: UI design through the GenomIUm method.

have been designed using the "Conceptual Framework" pattern. This pattern suggests that the UIs should share the same layout. The defined layout consists of three sectors: a heading, a body and a footer.

In step 3, *Content design*, the design patterns that compose each UI have been selected. Most of them pertain to the "Genomic Patterns" category, which addresses how to show and interact with genomic-related content. Table 1 describes the selected patterns for each UI.

In Step 4, *Refinement*, the selected patterns have been adapted to the specific particularities of the data to be displayed as well as the identified task that they solve. Step 4 in Figure 3 shows the refined *Visualization UI*. Only the refinement of the Visualization UI will be addressed due to space limitations. The re-

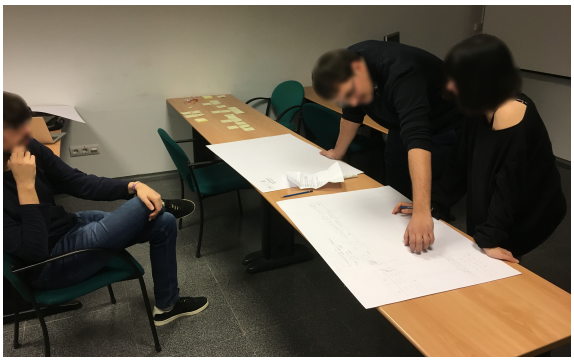

Figure 4: Geneticists and designers working together in a Card Sorting Session.

Table 1: UI patterns used in the Conceptual Design of the UIs.

Id	Pattern	Applied to
		Variety Selection UI
1	Set Operation	Define genomic data groups and compare them.
		Filter UI
2	Tabs	Separate the content into sections that can be accessed using a flat navigation (Toxboe, 2007)
3	Genetic Filter	Filter the variations by their type
4	Genetic Filter	Filter the variations by their frequency of appearance
5	Genetic Filter	Filter the variations by their quality
6	Genetic Filter	Filter the variations by their annotated impact
7	Genetic Filter	Filter the variations by their genomic region
8	Genetic Filter	Filter the variations by their allelic balance
9	Genetic Filter	Filter the variations by their position
10	Genetic Filter	Filter the variations by their effect over protein aspects
		Visualization UI
11	Chart	Show the number of genetic variations identified
12	Ideogram	Show the chromosome set
13	Chart	Show the distribution of genetic variations on each chromosome of the chromosome set
14	Ideogram	Show the detail of the selected chromosome in pattern 3
15	Chart	Show the distribution of genetic variations along the selected chromosome
16	Chart	Show the genetic variations distribution by varieties
17	Chart	Show the genetic variations distribution by scaffolds
18	Chart	Show the genetic variations distribution by Impact Annotation
19	Chart	Show the genetic variations distribution by Gene Ontology type
20	Chart	Show the genetic variations distribution by Enzyme type
21	Hidden Column	List the genetic variations involved in the overview visualizations (patterns 2 to 11)
		Present in the three UIs
22	Stepper	Guide users through the genetic analysis process

sulting Visualization UI gives the reader a clear idea of how the refinement process works.

The corresponding CD of the three UIs have been iteratively validated by bioinformaticians who provided valuable feedback to improve the UI designs. The refined CDs have been implemented with standard web technologies. More specifically, we designed the UI using the Angular framework[2], and utilized a set of open source libraries to implement the UI paterns (i.e., angular2-charts.js[3], ideogram.js[4], and agGrid[5]). These libraries offer angular-specific implementations that allows to include them in the project easily. Table 2 indicates the framework or library that implement each of the selected UI patterns.

Table 2: Frameworks and libraries used to implements the UI patterns used in the Conceptual Design of the UIs.

Id(s)	Pattern	Implemented with	License
		Variety Selection UI	
1	Set Operation	Angular	MIT
		Filter UI	
2	Tabs	Angular	MIT
3,4,5,6,7,8,9,10	Genetic Filter	Angular	MIT
		Visualization UI	
11	Chart	Angular	MIT
12,14	Ideogram	Ideogram.js	CC0 1.0 Universal
13,15,16,17,18,19,20	Chart	angular2-chartjs	MIT
21	Hidden Column	agGrid	MIT
		Present in the three UIs	
22	Stepper	Angular	MIT

Figure 5 illustrates the final implementation of the Visualization UI. Each pattern has been labelled according to the CD of the step four in Fig. 3.

5 VALIDATION

To validate the refined UIs, we have evaluated them by obtaining user feedback to confirm whether the UI design is in line with the user's needs. To do that, we have applied the User Interview technique which emphasises gathering information in an agile way rather than exhaustively documenting it (Preece et al., 2015). The evaluation process consisted of two steps: i) users are observed performing a set of previously defined tasks within their working environment using the refined UIs, and ii) users are interviewed to capture their impressions regarding their experience using the refined UIs.

5.1 First Step

A specific genomic analysis exercise has been defined to observe how domain experts use the application and interact with the developed UIs. This exercise consists of identifying meaningful variations by comparing two groups of varieties. One group contains four clementine varieties while the other contains four lemon varieties. Users must define several filters and interact with the result to identify the meaningful variations. Observing how domain experts performed the

[2]https://angular.io/

[3]https://github.com/emn178/angular2-chartjs

[4]https://github.com/eweitz/ideogram

[5]https://github.com/ag-grid/ag-grid

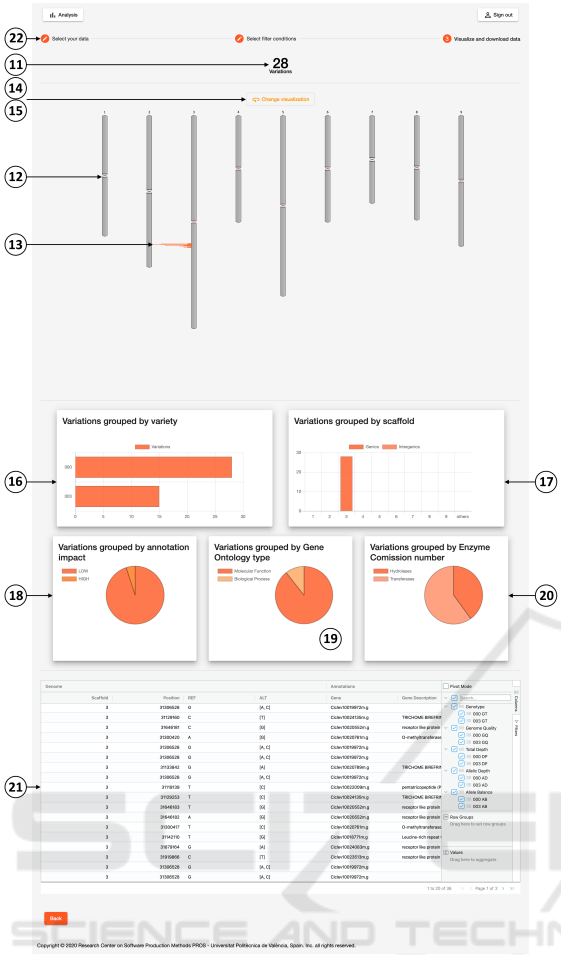

Figure 5: Implementation of Visualisation UI.

exercise allowed us to assess the well-operation of the application and to analyze the obtained results.

5.2 Second Step

We interviewed domain experts to know their opinion regarding the use of the application and its UI. The feedback obtained is summarized in three relevant points:

1. *The Application Provides Easy Guidance.*- They explained that the steps to follow in the analysis is easily described by the sequence of UIs (i.e. variety selection UI, filter UI, visualization UI) improving the way the analysis is performed.
2. *It Is Easy to Learn.*- Users found intuitive to manage the application because the tasks to perform with the UIs match real work environment tasks. They perform the analysis of the genetic variations with minimum technological support. Besides, genomic terminology used in the UIs is familiar to the user.
3. *Greater Access.*- Users mentioned that the proposed UI expands the possibility of access to data to experts and novices bioinformaticians. In the traditional process, analyzing the data was limited to expert bioinformaticians with high computer skills.

In general, domain experts reported a positive use of the implemented solution. The reason is that the provided tool allowed them to reduce execution time easily and intuitively. Other solutions either decreased execution time too little or were too complicated and required a large amount of time to learn to use them. Nevertheless, they mentioned that adopting the tool takes a small amount of time to get used to it and change their mental model; but they stated that the benefits of using the tool outweighed the cost of adopting it.

In conclusion, it is an undeniable fact that the automatizing of the genomic analysis process produces greater satisfaction and benefits than performing it manually. However, how easy it is to use the application that automates the process depends largely on how easy it is to use its UI. The bioinformaticians opinion shows that the designed UI eases the use of the application and reduces the complexity of performing the genomic analysis. The validation reported encouraging findings but the results should be understood under the conditions of the evaluation. Our next step in this line is to carry out more empirical evaluations that reinforce the results obtained.

6 CONCLUSIONS

UIs are crucial to manage data and extract knowledge from it. Its design and development require proper attention as good designed UIs can have a huge impact in performing these tasks. Unfortunately, genomic applications are unintuitive, complex and overly verbose because their UIs are poorly designed. Consequently, learning to use them is difficult and tedious, which reduces knowledge extraction from genomic data.

This paper emphasizes the use of UCD as an appropriate approach to design genomic UIs. We report the design and implementation of genomic UIs in a real-word use case by applying UCD techniques and GenomIUm, a POD based method where each UI is composed of a set of UI patterns. Firstly, the relevant tasks of the use case have been identified and studied through UCD techniques. Secondly, the UIs have been designed according the GenomIUm method by

selecting the most appropriate patterns for each identified task. Thirdly, the designed UIs have been evaluated by bioinformaticians.

Complementing UCD techniques with the support of a pattern-based method (i.e. GenomIUm) to design UIs provides greater benefits. While UCD techniques allows to research the users and to specify the real user tasks, the method guides the design and implementation of the UI based on the user tasks. Composing UIs with widely used design patterns (provided by GenomIUm) makes them familiar and consequently easy for bioinformaticians to use.

The UCD approach together with the GenomIUm method allowed us to generate high-quality UIs. Bioinformaticians reported to be satisfied with them as it allowed them to improve knowledge extraction and data management processes by i) automating the process, ii) providing an intuitive guideline to bioinformaticians, iii) allowing to deal with huge amount of data that is complex in nature and iv) removing the need of having high computer skills.

Future work includes, on the one hand, to carry out a broader, more empirical user evaluation. This evaluation should measure the increase of the user's performance when using pattern-based UIs. On the other hand, the continuous improvement of the GenomIUm method with the inclusion of new UI patterns.

ACKNOWLEDGEMENTS

The authors would like to thank the members of the PROS Research Center Genome group for fruitful discussions regarding the application of Conceptual Modeling in the medical field. This work has been developed with the financial support of the Spanish State Research Agency and the Generalitat Valenciana under the projects TIN2016-80811-P and PROMETEO/2018/176 and co-financed with ERDF. Work at IVIA is funding by the Ministerio de Ciencia, Innovación y Universidades (Spain) trough grant RTI2018-097790-R-100 and by the Instituto Valenciano de Investigaciones Agrarias (Spain), through grants 51915 and 52002.

REFERENCES

Al-Ageel, N., Al-Wabil, A., Badr, G., and AlOmar, N. (2015). Human Factors in the Design and Evaluation of Bioinformatics Tools. *Procedia Manufacturing*, 3:2003–2010.

Bolchini, D., Finkelstein, A., Perrone, V., and Nagl, S. (2009). Better bioinformatics through usability analysis. *Bioinformatics*, 25(3):406–412.

Carpenter, A. E., Kamentsky, L., and Eliceiri, K. W. (2012). A call for bioimaging software usability.

Chilana, P. K., Wobbrock, J. O., and Ko, A. J. (2010). Understanding usability practices in complex domains. In *Conference on Human Factors in Computing Systems - Proceedings*, volume 4, pages 2337–2346, New York, New York, USA. ACM Press.

Cingolani, P., Platts, A., Coon, M., Nguyen, T., Wang, L., Land, S., Lu, X., and Ruden, D. (2012). A program for annotating and predicting the effects of single nucleotide polymorphisms, snpeff: Snps in the genome of drosophila melanogaster strain w1118; iso-2; iso-3. *Fly*, 6(2):80–92.

Clément, L., Emeric, D., J, G. B., Laurent, M., David, L., Eivind, H., and Kristian, V. (2018). A data-supported history of bioinformatics tools. *arXiv preprint arXiv:1807.06808*.

de Matos, P., Cham, J. A., Cao, H., Alcántara, R., Rowland, F., Lopez, R., and Steinbeck, C. (2013). The Enzyme Portal: A case study in applying user-centred design methods in bioinformatics. *BMC Bioinformatics*, 14.

Galperin, M. Y. (2008). The molecular biology database collection: 2008 update. *Nucleic Acids Research*, 36(SUPPL. 1):D2.

Goodwin, S., McPherson, J. D., and McCombie, W. R. (2016). Coming of age: Ten years of next-generation sequencing technologies. *Nature Reviews Genetics*, 17(6):333–351.

Iñiguez-Jarrin, C. (2019). *GenomIUm: A Pattern Based Method for Designing User Interfaces for Genomic Data Access*. PhD thesis, Universitat Politècnica de València.

Jaspers, M. W. (2009). A comparison of usability methods for testing interactive health technologies: Methodological aspects and empirical evidence. *International Journal of Medical Informatics*, 78(5):340–353.

Javahery, H. and Seffah, A. (2002). A Model for Usability Pattern-Oriented Design. In *Proceedings of the First International Workshop on Task Models and Diagrams for User Interface Design*, TAMODIA '02, pages 104–110. INFOREC Publishing House Bucharest.

Javahery, H., Seffah, A., and Radhakrishnan, T. (2004). Beyond Power: Making Bioinformatics Tools User-centered. *Commun. ACM*, 47(11):58–63.

Mardis, E. R. (2011). A decade's perspective on DNA sequencing technology. *Nature*, 470(7333):198–203.

Paternò, F. (2003). ConcurTaskTrees: An Engineered Notation for Task Models. *The Handbook of Task Analysis for Human-Computer Interaction*, pages 483–503.

Pavelin, K., Cham, J. A., de Matos, P., Brooksbank, C., Cameron, G., and Steinbeck, C. (2012). Bioinformatics meets user-centred design: A perspective. *PLoS Computational Biology*, 8(7):e1002554.

Preece, J., Rogers, Y., and Sharp, H. (2015). *Interaction design: beyond human-computer interaction*. John Wiley & Sons Inc, 4 edition.

Rimmer, J. (2004). Improving software environments through usability and interaction design.

Rutherford, P., Abell, W., Churcher, C., McKinnon, A., and McCallum, J. (2010). Usability of navigation tools for browsing genetic sequences. In *Conferences in Research and Practice in Information Technology Series*, volume 106, pages 33–41.

Stephens, Z. D. et al. (2015). Big data: Astronomical or genomical? *PLoS Biology*, 13(7):e1002195.

Stevens, R., Goble, C., Baker, P., and Brass, A. (2001). A classification of tasks in bioinformatics. *Bioinformatics*, 17(2):180–188.

Sutcliffe, A., Thew, S., De Bruijn, O., Buchan, I., Jarvis, P., McNaught, J., and Procter, R. (2010). User engagement by user-centred design in e-Health. In *Philosophical Transactions of the Royal Society A: Mathematical, Physical and Engineering Sciences*, volume 368, pages 4209–4224. Royal Society.

Svanæs, D., Das, A., and Alsos, O. A. (2008). The contextual nature of usability and its relevance to medical informatics. In *Studies in Health Technology and Informatics*, volume 136, pages 541–546.

Toxboe, A. (2007). User interface design pattern library.

Tran, D., Dubay, C., Gorman, P., and Hersh, W. (2004). Applying task analysis to describe and facilitate bioinformatics tasks. In *Studies in Health Technology and Informatics*, volume 107, pages 818–822.

Valentin, F., Squizzato, S., Goujon, M., McWilliam, H., Paern, J., and Lopez, R. (2010). Fast and efficient searching of biological data resources-using EB-eye. *Briefings in Bioinformatics*, 11(4):375–384.

Wu, G. A. et al. (2014). Sequencing of diverse mandarin, pummelo and orange genomes reveals complex history of admixture during citrus domestication. *Nature Biotechnology*, 32(7):656–662.

Wu, G. A. et al. (2018). Genomics of the origin and evolution of Citrus. *Nature*, 554(7692):311–316.