Rodrigo, M. M. T. et al. (Eds.) (2021). Proceedings of the 29th International Conference on Computers in Education. Asia-Pacific Society for Computers in Education

Karyotype: An Interactive Learning Environment for Reasoning and Sense Making in Genetics through a Case-based Approach

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Abstract: Reasoning and sense-making has been proved to be difficult for learners. Connecting genetic disorders with their underlying chromosomal aberrations requires reasoning and sense-making based on the clinical symptoms. In traditional instructional methods, learners encounter the required concepts and scientific processes in different grades and across different settings – theory classes, labs, tutorials, and most tests assess learners' ability to remember the facts. This leads to a focus on vocabulary and memorization, rather than on scientific reasoning and making connections across different concepts and processes. To address this gap, we have developed *Karyotype*: a web-browser based learning environment where learners assume the role of a geneticist and solve the cases of chromosomal disorders. The learning environment is based on a case-based reasoning approach where the learner acts as a problem solver. S/he is given a set of symptoms and is asked to explain them and suggest a diagnosis. In this paper, we present the theoretical basis and design of *Karyotype*. We report an exploratory study in which we investigated learning of problem-solving skills and perceptions of usability and usefulness of techno-pedagogical features of *Karyotype*. The results indicate that learning activities and scaffolds in *Karyotype* are helpful for learning reasoning and sense-making skills.

Keywords: Interactive content, chromosomal disorders, reasoning, sense-making, case-based approach

1. Introduction

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. One such type is chromosomal disorders, caused due to abnormalities such as a change in the number or structure of entire chromosomes, or a specific part of the chromosome carrying a particular gene. Students pursuing bioscience majors encounter these concepts to varying levels of breadth and depth in their undergraduate and postgraduate curriculum. They learn about genetic mutations that cause disorders, their symptoms, and their effects as well as the diagnostic methods and treatments. However, often they learn these topics in the form of disjoint facts, which they perceive as a memorization activity. Students fail to make the connection between the defects at the chromosomal level and their corresponding effect on the features displayed by an individual. Making these connections requires reasoning in terms of physiological states, complaints, and symptoms. Typical curricula cover the concepts and procedures required for diagnosis in silos in theory classes, practical labs, and tutorial sessions. What gets missed is the emphasis to facilitate learners through the reasoning process and making explicit connections across the concepts.

An important process in diagnosing genetic disorders is karyotyping. The karyotyping activity has been traditionally conducted as a pen and paper exercise where learners are provided with a print-out of the chromosome spread. Learners are expected to arrange the chromosomes in a particular order as per their size, the position of the centromere, and the length of p and q arms of a chromosome. This is done by matching the homologous pairs to complete the karyotype which is a map of human chromosomes. After completion of the karyotype, the anomaly can be detected as any additional/missing chromosome. An extension of the traditional pen and paper activity includes karyotyping kits developed by the Science Source (The Science Source SE, 2021), Thermo Fisher

Scientific (Thermo Fisher Scientific, 2021), and Carolina (Carolina, 2021). While these products are in the form of tangible objects available offline, there are a few efforts made to bring the interactivity elements via online simulations (Labster Cytogenetic, 2021), virtual labs (Virtual cell biology lab, 2021), and web-based approaches (The Biology Project, 2021; Learn Genetics, 2021; Training Karyotypes, 2021). These systems provide stand-alone activities that make use of interactivity for the arrangement of chromosomes to complete a karyotype. However, this approach does not focus on learners making connections between the cause and effect associated with an abnormal chromosomal condition. Instructional strategies combined with interactive visual teaching resources would aid students in obtaining a large amount of knowledge and remembering more ideas (Riyanto, Amin, Suwono & Lestari 2020). Few systems provide example cases for the learners before they move on to the karyotyping activity.

There is a need for a learning environment that can provide hands-on experience of the complete process followed for the diagnosis of disorders. Technology-enhanced learning environments can be used for facilitating case-based reasoning by providing overall structure to the interactive learning activities, immediate feedback, scaffolds in form of reflective and evaluative question prompts, and so on (Deep, Murthy & Bhat, 2020). We propose an online learning environment *Karyotype* that provides learners with a series of interactive learning activities situated in the context of specific cases of disorders wherein learners diagnose the disorder based on the underlying genetic condition, while reasoning through different phases of inquiry.

In this paper, we describe the theoretical basis and design of *Karyotype*, and report a pilot study with 5 bioscience majors' learners in the context of chromosomal disorders. The two research objectives of this study are to examine the effectiveness of the scaffolds in the system to help learners in reasoning and sense making, and to understand learners' perception about learning activities.

2. Theoretical Basis

Sense-making about a phenomenon involves collecting observations, analysing the data and constructing interpretations. Sense-making and reasoning are closely associated in order to strengthen the interpretations. Stories are the oldest and most natural form of sense making as they allow explanation and interpretation. It helps to develop empathy towards the central character of the story, which acts as a way to understand a situation better (Herreid, 2007; Jonassen and Hernandez-Serrano, 2002; Centre for teaching and learning 2021). Exposing the learners to different cases or stories while solving problems, provides them an opportunity to reflect in action (Jonassen and Hernandez-Serrano, 2002). Combining the two together is the cased-based learning approach which presents stories in the form of cases to be solved. Case-Based Reasoning (CBR) proposes a method in which students learn by doing problem solving and other activities that pique their interest and allow them to apply what they've learned in a way that provides immediate feedback (Kolodner et al, 2003). CBR has been used in medicine for diagnosis as the methodology of CBR systems closely resembles the thought processes of a physician. This reasoning includes cognitive activities similar to sense making, like gathering information, recognition of patterns, solving problems, and decision making (Choudhury & Ara, 2016). In CBR, the remembered cases are used as a means of efficient problem-solving. Here elicitation becomes a task of gathering case histories and implementation is reduced to identifying significant features that describe a case (Watson and Marrir, 1994).

3. Design and Development of *Karyotype*

Karyotype is an online, self-paced, task-oriented learning environment. The target learners of *Karyotype* are biology majors' students. Educators propagating basic science can make use of *Karyotype* as an instructional system. Additionally, *Karyotype* can cater to learners across the disciplines and age groups, who are interested in acquiring knowledge about the genetic material and its impact on our lives in general. The learning environment has a series of learning activities that helps the learners to have a complete walkthrough of the process followed for clinical diagnosis of chromosomal abnormalities. The system makes use of interactive elements to enhance learners' engagement with the content and

provides scaffolds for the learners to help them make progress in the learning activities that are based on various phases of an inquiry cycle which guide learners to achieve the intended learning outcomes. An interactive video narrative presented to the learners in the form of a case history of a patient (Fig 1, top left), acts as an anchor providing an authentic and engaging narration to help learners understand and empathize with the patients' story. Learners have the autonomy to choose any case of their interest based on the case briefing provided. Reflection spots within the video (Fig 1, top right) are used as scaffolds to help in the better understanding of the problem context and identification of the symptoms associated with a disorder. Hints are provided to help learners choose appropriate explanations from the given set of options. Constructive feedback is given to scaffold the learners in concluding the reflection activity. Drag and drop activity along with hints and feedback (Fig 1, bottom left) to move the chromosomes in order to complete a karyotype, helps the learners in analysing and interpreting the underlying genetic condition. This allows identifying the anomaly in a given karyotype. The help and information prompts (Fig 1, bottom right) associated with interactive chromosome images are used to scaffold the learners to make connections between the visible symptoms and invisible genetic conditions. Interactive images act as redundant scaffolds providing more opportunities for learners to access additional information as and when required. Learners are supposed to prepare a diagnosis report based on their observations and the reference material provided. A look-up table is provided to scaffold collating observations and results from previous phases of inquiry for making a final prediction about the disorder. The report needs to have reasons and justifications for the choice made by a learner. The purpose of this is to help improve the reasoning skills of learners (Kolodner et al, 2003). Using the correct set of reasoning would help in making a correct diagnosis, thereby aiding the problem-solving process.

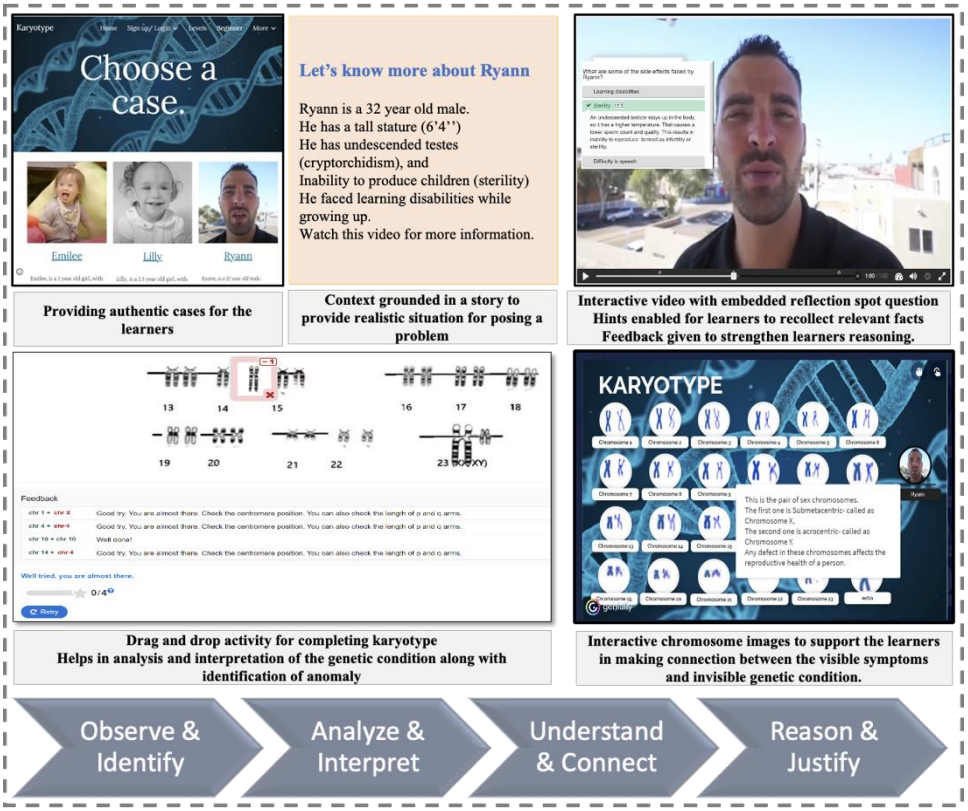

Figure 1. Learning Activities in Karyotype.

A few examples of chromosomal disorders included in *Karyotype* are Turner's syndrome, Larsen syndrome, and Jacobsen syndromes. The user interface of *Karyotype* is designed and implemented with Google sites, which is an open-source toolkit. The learning activities of *Karyotype* have been designed in H5P and genially. H5P is an open-source, HTML5 toolkit to develop interactive content. *Karyotype* can be accessed through a standard web browser using any device.

4. Study Design

The purpose of this pilot study was to understand the role of the pedagogical design features of *Karyotype* through participants' experience, performance and perceptions, and inform redesign. The participants of this study were 5 bioscience majors' students from one of the colleges in Kerala, India. In this study, we chose chromosomal disorders as the context covered in the learning material. Problem-solving in this topic requires the students to understand the context of the problem, make relevant observations regarding the case, perform basic tests for diagnosis and come up with a reasoning and justification for a possible diagnosis associated with the given task.

This study was conducted as a part of a workshop for bio-science learners. It was conducted online in the presence of the instructor in a supervised setting. Google Meet was used as the online video conferencing platform for the synchronous session and post workshop interviews. The *Karyotype* learning environment was accessed as a Google Site using the standard web browser for capturing data of the learning gains on case-based reasoning and their perception of scaffolds present in the LE. The study had five steps - Registration and self-perception survey, Pre-Test, Interaction with the LE, Post-test and post workshop interviews. The registration form recorded participants personal and academic information along with their confidence (on a likert scale of 1-5, ranging from low to high) regarding the knowledge of chromosomal abnormalities, preparing a karyotype, analyzing and interpreting a given karyotype, and, reasoning about the causes and symptoms associated with a chromosomal disorder.

There are 4 data sources in the study which includes Pre-test, reports of *Karyotype* learning activities, report from post-test, and interviews. Reports of learning activities include notes and observations made by learners and their diagnosis. The pre-test consisted of two questions to understand how learners make sense of the given problem and the role of providing a context. Each participant solved 3 cases during their interaction with *Karyotype*. After going through a series of learning activities (Fig.1), students generated diagnosis reports which were to understand the role of the scaffolds in the diagnosis process. In the post test, these scaffolds were withdrawn. Participants' responses from the artefacts submitted in the form of a diagnosis report were analysed on three criteria related to sense making: C1 – Making explicit and relevant clinical observations, C2 – Connecting the observable clinical symptoms with the chromosomal condition and C3– Explaining how chromosomal changes lead to clinical symptoms. A 3-point rubric (0-missing, 1-needs work, 2- adequate) was used to analyse the data from pre-test, final diagnosis report and post-test. The semi-structured stimulated recall interviews were conducted to get insights regarding participants' perception about the techno-pedagogical design and learning activities in their process of diagnosis and sense making. Participants were probed about their experience of interacting with *Karyotype* and how they made use of the learning activities to solve the given case. 30 instances of participants' use of case-based learning strategies and *Karyotype* design elements were identified.

5. Findings

5.1 Performance, Usability and Usefulness of Karyotype

Table 1 represents the average performance of participants for the three criteria in the pre-test, within the activities of the intervention and in the post-test.

Table 1. *Participants (P) average scores*

P	Overall			C1			C2			C3		
	Pre	Int.	Post	Pre	Int.	Post	Pre	Int.	Post	Pre	Int.	Post
S1	0.66	1.66	1	0	2	0	1	2	2	1	1	1
S2	1	1.33	1.33	1	2	2	1	1	1	1	1	1

S3	0.66	1.66	1.66	0	2	2	1	2	2	1	1	1
S4	1.33	1.66	1.33	2	2	1	1	2	2	1	1	1
S5	1	1.33	NA*	1	2	NA*	1	1	NA*	1	1	NA*

*Did not attempt due to technical reasons

The majority of learners were found to make explicit and relevant clinical observations. A sample observation by a participant is as follows: *"Emile is suffering from a genetic disorder. The symptoms shown by her are a sudden drop in blood sugar level so she is admitted in NICU and fed through nose. She has been given psychological support to avoid mental breakdown."* However, S1 missed noting down the observations in the post test which could be due to the absence of scaffolds in the post test. Participants found the interactive videos with reflection spots to be useful for making clinical observations. S2: "*There are many questions in between the video which helps to summarize the symptoms of the syndrome. It was very helpful.*" It was found that while making notes and observations about a case, participants made an emotional connection and expressed empathy towards the patient. S4: *"Nora's parents were disappointed when the doctors say there is no hope for the child and if she was born, they can't say how long she will live. When she was born, they were very happy to see her. Now they are very happy about her performance. She can smile, sing. And her father was disappointed about her cleft lips."*

Learners' performance in connecting the observable symptoms with the chromosomal condition (Table 1) improved from "needs work" level in pre-test to "adequate" in learning activities and post-test. This performance remained consistent in the post test after removal of scaffolds suggesting that learners can become a more efficient reasoner by remembering old solutions and adapting them rather than having to derive answers from scratch each time (Kolodner, 1992). *S3: "Hints are always useful. Okay. But towards the end, it's not much difficult for me."* This is a key feature of the case-based reasoning approach considered as a methodology for both reasoning and learning. Participants found the look-up table useful for moving towards the correct diagnosis by matching the symptoms and underlying chromosomal condition. *S1: "Lookup table was required, because while considering this patient's symptoms, we have some doubts that it may be this. So, by looking at this table, we can confirm it. It is this disease." "By seeing the video, we can understand their phenotypic characteristics. And then by the lookup table, we can understand the genotype."* However, the performance in explaining how chromosomal changes lead to clinical symptoms (Table 1) remained constant with no significant increase or decrease suggesting that spending more time with the LE and exposure to more cases might be required.

6. Discussion and Future Work

The techno-pedagogical design elements used in the LE as per the guidelines about the role of scaffolds in software assisted learning (Quintana et al, 2004), when mapped with their intended purpose with respect to the learning outcomes for learners were found to be consistent with the findings from the interviews and artefacts. Making clinical observations and noticing what is important about scientific situations requires substantial conceptual domain-specific knowledge, which novice learners may lack. It's beneficial to incorporate expert input to assist learners in making connections to things they are familiar with while also challenging them to comprehend new phenomena. Participants' responses about the interactive videos and hints/feedback provided during the drag and drop activity, suggest that having such visual representation helps learners understand the problem context better, aid observations and problem solving. Engaging in reflective self-assessment can increase learners' understanding about the content and the inquiry process. It is observed that sometimes learners conceive opportunities for articulation and reflection as mere blank fields to be filled in and miss out on productive reflection. Scaffolds can help by focusing learners' attention on making strategic decisions they might otherwise avoid. Interactive chromosome images provide visual representations that may be examined to expose data's underlying qualities. Look-up table helps engage in sense-making practices while reflecting on

disciplinary strategies. The report template provides a mechanism for recording observations, findings, or ideas during the diagnosis process. It also helps the learners to express their thinking in ways that highlight important disciplinary ideas. Our next development work includes analysis of the users' feedback to modify and implement additional functionalities in the learning environment, and conducting further user studies to understand how learners navigate through the process of reasoning and sense-making.

Acknowledgements

We thank Next Education Labs, Educational Technology, IIT Bombay for the support provided in developing *Karyotype*. We thank all the people whose stories and videos were used for creating the learning activities. We thank Prof. Praveen Prasad and our participants from SN College, Nattika for helping us with this research study and Mohd. Saif for the beta testing of the system.

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