Tittle:

Genomic medicine in motion - Expanding genomic medicine awareness in Latin America using dynamic motion graphics capsules for patients and their families.

Contact Information:

Eduardo Esteban Pérez Palma, PhD / eduardoperez@udd.cl / +56 950 731 971
Avenida Apoquindo 6797, Las Condes, 7570003, Región Metropolitana de Santiago, Chile.

Introduction:

Recent developments in genomic research alongside the massive expansion of clinical genetic testing has produced vast amounts of biomedical knowledge, novel resources, and therapies[1,2]. Today, genetic testing and the clinical genetic counseling profession have become an essential part in the diagnosis and management of all rare and many common genetic disorders[3,4]. However, genomic medicine implementation in Latin America is far behind northern countries. Having similar incidences for mendelian genetic disorders and an understudied pool of common genetic variation conferring risk to common disorders[5,6], the need for genomic medicine implementation is equal. Naturally, thousands of genetic tests are carried out on Latin American patients. However, Latin American clinical genetic testing is mostly done abroad, and the genetic counseling profession remains unrecognized with only a few certified professionals in active patient care[3,7]. Further, the number of trained medical geneticists are far below the number recommended[7], leading to long waiting times and low-quality to non-existing genetic counseling.

Education is among the main functions of genetic counseling. Helping patients and their families to understand basic but complex genomic medicine concepts such as inheritance, testing, genotype-phenotype correlations, management, resources and research[3] can significantly improve their quality of life[8] and even accelerate applications[9]. Here, the educational needs of the community affected by genetic disorders are diverse and specific. The urgency for information does not match with a formal genetic course. Available genomic medicine educational resources are mostly in English and can be overwhelming to the lay person. Thus, we recognize a need for an adapted set of basic genomic medicine concepts. Here, videos, animations and motion graphics can aid in the education of complex medical concepts[9,10]. Thus, under the hypothesis that: "*Dynamic motion graphics capsules have a positive effect on the Latin American community affected by genetic disorders*" in this project, I propose to create series of 18 motion graphic capsules for Latin American patients and their families to educate themselves about core genomic medicine concepts.

Vision:

We will cover major genetic disorder types, modes of inheritance, types of genetic testing, patient management, resources available, and current Latin American research opportunities. Capsules will be hosted in an interactive webserver that will assess the user needs through a brief set of multiple choices ("Type of disorder", "Type of test", "Pathogenic or VUS"). Webserver will be developed with the Shiny R framework following previous templated developed by our group[11,12]. Capsules will have a dynamic nature and will be connected according to the needs of the user. We will produce two types of capsules: 1) general capsules containing information relevant to all users (e.g., "Genomic Medicine", "Situation in Latin America", "Genetics"); and 2) specific capsules containing information relevant only to a subset of users. Regardless of user choice all capsules will be available to the public. The full description of the 18 capsules in shown in Figure 1. Finally, to quantitative evaluate the benefits of using motion graphics capsules in the community we will invite participants to participate in a non-mandatory additional survey contained in the same platform. To visualize our concept, we have produced a teaser capsule in the following link describing "Rare diseases": https://vimeo.com/589633753/c6be6b83df.

Aims:

1. To create a series of 18 motion graphic capsules for Latin American patients and their families to educate themselves about genomic medicine according to their own needs.
2. To evaluate the overall benefit of using motion graphic capsules in the Latin American community affected by genomic disorders.

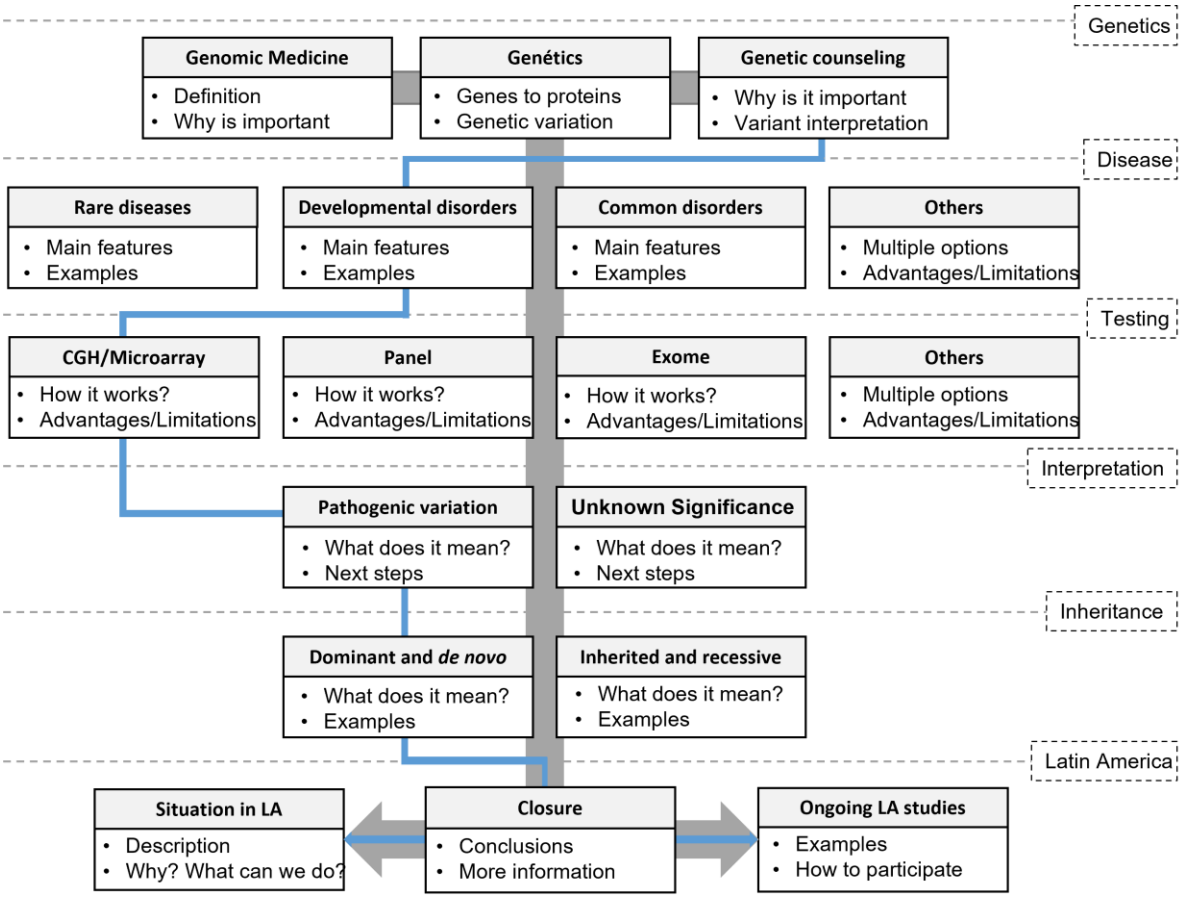

Figure 1. Strategy. We will create 18 capsules distributed in 6 levels: Genetic, disease, testing, interpretation, inheritance, and Latin America (LA). Genetics (top) and LA (bottom) are general capsules intended for all users. Remaining capsules will be displayed dynamically according to the users' educational needs. Grey arrows show the progressive order of visualization. Users can follow from top to bottom several paths. An example capsule path is shown with the blue line.

Impact:

Our project will contribute to bridge the knowledge gap between the basic scientific concepts in the genomic medicine field and the community affected by genetic disorders in Latin America. We will provide to Spanish and Portuguese speakers a novel and easy-to-digest access to key genetic concepts. Thus, our project will have a direct impact to patients and their families affected by genetic disorders contributing to lower the psychological burden associated with the lack of information and appropriated genetic counseling. Further, will update and connect interested parties with the latest regional developments in genomic medicine.

Executive plan:

Quarterly plan.	Q1	Q2	Q2	Q2
Aim 1. To create a series of motion graphic capsules for Latin American patients and their families to educate themselves about genomic medicine according to their own needs.				
-Interactive webserver development.				
-Pre-production (design, motion plan, concept definition, scripts, etc.).				
-Production, Post-production (animation, music, voice overs, etc.).				
-Release.				
Aim 2. To evaluate the overall benefit of using motion graphic capsules in the Latin American community affected by genomic disorders.				
-Survey design.				
-Survey application.				
-Survey analysis.				
-Manuscript preparation.				

G2MC Partner.

Dr. Gabriela Repetto is a Clinical Geneticist and Director of the Center for Genetics and Genomics at Facultad de Medicina Clinica Alemana Universidad del Desarrollo in Santiago, Chile, a leading clinical, teaching and research interdisciplinary center in Medical Genetics and Genomics. Dr. Gabriela Repetto is an active member of G2MC.

Goals.

1.- Produce 18 motion graphic capsules described in Figure 1.
2.- Develop an interactive webserver to host all capsules and specific capsules view-paths.
3.- Survey users to quantitively evaluate the overall benefits of the tool.
4.- Analyze and report results to the community.

Other organizations:

Project management and budget administración: Centro de genética y Genómica, Facultad de Medicina Clinica Alemana, Universidad del Desarrollo.

Future funding:

With the successful implementation of our project, we distinguish at least two branches with potential to obtain future funding. First, platform expansion. To increase the number of topics, length and depth covered by our capsules. Such an expansion will contribute to increase awareness of genomic medicine in the general population and provide specific information to more patients. Second, integrate patient recruitment. The communication of ongoing clinical trials and medical research is challenging. An appropriate communication is essential to reduce some of the barriers associated with poor patients' enrollment in clinical trials and with patients' uninformed consent or uninformed refusal.

References

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Budget:

Materials and supplies			
Animated capsules (n=18)			**$ 24,300**
Justification: We will create 18 motion graphics 2D video capsules of approximately 1 minute duration each (depending on each script). We will use the Infographics & Iconographics style. The digital animator of choice is Diego Perez (https://vimeo.com/user110886899) who developed the teaser and quote prices detailed below in USD:			
Pre-Production	Script assistance	$	40
	Storyboard	$	70
	Style frame	$	50
Production	Design & Illustration	$	350
	Animation	$	400
	Editing	$	100
	Music Selection	$	50
	Voiceover	$	240
Post-Production	Render	$	50
	Total price per animated capsule	$	1,350
Personnel			
Bioinformatic Support: To be hired			**$ 7,800**
Justification: With 10 hours per week (US $15 dollars per hour in 52 weeks) of dedication to this project the Bioinformatic support will contribute to organize scientific content of the scripts, scan the current state of genomic medicine in Latin America and help to develop the interactive web application.			
Principal Investigator: Eduardo Pérez, PhD			**$ 6,240**
Justification: With 6 hours per week (US $20 dollars per hour in 52 weeks) of dedication the PI will supervise the whole project. With extensive experience in the genetic disorders, variant interpretation and web application development Dr Eduardo Perez will ensure accurate script development.			
Travel and meeting support			
Support to travel to G2MC meeting in 2022			**$ 2,500**
Justification: The project will fund travel of one member of the team to the G2MC meeting in 2022 to show the current state of the project and obtain feedback from the stakeholders community			
Support for local/country involvement			
Administrative cost at the hosting institution			**$ 5,000**
Justification: The hosting and collaborating institution will be the Centro de Genetica y Genomica de la universidad del Desarrollo. Here, university will support the PI in the administration of the project and managing budget. It will provide office and meeting space among other non-monetary support (Wi-Fi, computers, etc.).			
Tower computer			**$3,000**
Justification: The project involves computational analysis, web development and animation rendering. A high-performance tower computer is needed. The computer will be added to the lab and contribute to research and education.			
Miscellaneous costs			
Cloud computing and webhosting			**$ 1,000**
Justification: The project plans to create and host a interactive webserver to display the capsules and survey. Based on PI previous experience, we will use gcloud services.			
		Total project:	**$49,840**