English

Website for information on rare diseases of genetic origin: first step for telegenetics in Minas Gerais, Brazil

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Abstract

Introduction: Telegenetics can ease the paradox of Medical Genetics in Brazil: high patient demand versus few specialized professionals. Method: Descriptive study of the implementation of a website with information about rare diseases for the population and health professionals. Results: Telegenetica-MG works since 03/01/2019 and had 176 accesses in 4 months. Conclusion: It is a step for telemedicine to be applied to genetics in Minas Gerais.
Keywords: Telemedicine, Genetics, Rare Diseases.

Resumen

Sitio web de información sobre enfermedades raras de origen genético: primer paso hacia la telegética en Minas Gerais, Brasil
Introducción: La telegenética puede aliviar la paradoja de la Genética Médica en Brasil: alta demanda de pacientes frente a pocos profesionales especializados. Método: Estudio descriptivo de la implementación de un sitio web con información sobre enfermedades raras para la población y los profesionales de la salud. Resultados: Telegenetica-MG ha estado funcionando desde el 01/03/2019 y tuvo 176 visitas en 4 meses. Conclusión: Es un paso hacia la aplicación de la telemedicina a la genética en Minas Gerais.
Palabras clave: Telemedicina, Genética, Enfermedades raras.

Resumo

Website para informação sobre doenças raras de origem genética: primeiro passo para telegenética em Minas Gerais, Brasil
Introdução: A telegenética pode amenizar o paradoxo da Genética Médica no Brasil: alta demanda de pacientes versus poucos profissionais especializados. Método: Estudo descritivo da implantação de uma página web com informações sobre doenças raras para a população e profissionais de saúde. Resultados: O Telegenetica-MG funciona desde 01/03/2019 e teve 176 acessos em 4 meses. Conclusão: É um passo para que a telemedicina seja aplicada à genética em Minas Gerais.
Palavras-chave: Telemedicina, Genética, Doenças Raras.

Introduction

Rare diseases are those that affect up to 1.3 out of every 2,000 individuals[1]. Although individually rare, as a group, they affect a significant percentage of the population. In Brazil, it is estimated that 13 million people are affected by diseases considered rare[1]. Among the rare diseases, 80% have genetic origin, such as congenital anomalies, intellectual disability and innate errors of metabolism.

The geneticist has a prominent role in the management of such diseases. Currently, there are less than 300 active geneticists in the area throughout the country, being the medical specialty with fewer professionals. On the other hand, the importance of genetic diseases has increased exponentially in recent years. Congenital defects passed from the fifth to the second cause of infant mortality between the years 1980 and 2000 in Brazil[2].

One of the possibilities to solve the impasse in which Medical Genetics is found in the Brazilian Unified Health System (SUS), with repressed demand and few specialized professionals, is the use of telegenetics, which has been studied in recent years[2,3,4].

In Minas Gerais, there are no services that offer telegenetics. To fill this gap, a website was developed, called TelegeneticaMG, containing information on rare diseases of genetic origin.

Method

This is a descriptive study of the implementation of a TelegeneticaMG website, developed in Java language and the data is stored in MySQL database. It is composed of two sections with restricted access: one for patients, their relatives and the general population, and the other, which includes specific information for professionals involved in the care of patients with rare diseases. To have access to the restricted area, the user must sign a specific Term of Free and Informed Consent for each category and fill out the registration form informing demographic data, such as gender, age, profession, place of work and reason for interest in rare diseases. The project was approved by the UFMG Research Ethics Committee and the Teaching and Research Management of the Clinical Hospital of UFMG.

The final version of the website is available for access since 03/01/2019, through the electronic address http://www.telegeneticamg.com.br.

Results and Discussion

TelegeneticaMG had 176 accesses (87 in the category "health professionals" and 89 in "patients, family and general public" in the period between 01/03/2019 and 01/07/2019.

In the category "health professionals", the majority of the participants were women (76%) and residents of the central macroregion of Minas Gerais. The most frequent professional area was the medical (49.4%), being the medical genetics the most frequent specialty (27.9), followed by pediatrics (25.6%) (Table 1 and graphic 1).

Table 1. Profile of the registered user (health professionals) of the TelegeneticaMG website

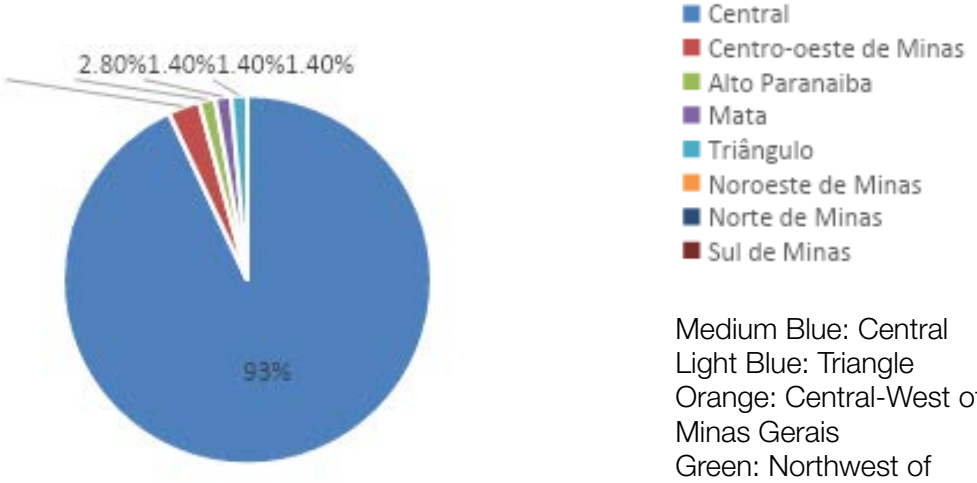
SEX			
Feminine		66 (75,9%)	
Masculine		21 (24,1%)	
Total		87 (100%)	
WHERE ONE RESIDES			
MG		71 (81,6%)	
SP		5 (5,75%)	
BA/RJ/DF		2 in each state (6,9%)	
MA/PR/PA		1 in each state (3,45%)	
Paraguay		1 (1,15%)	
NS*		1 (1,15%)	
Total			
HAS THE DIAGNOSIS OF A GENETIC DISEASE			
Yes		ACCOMPANIES WITH A GENETICIST	
	3 (3,45%)	Yes	0 (0%)
		No	3 (100%)
No	84 (96,55%)		
Total	87 (100%)		

ACCOMPANIES WITH A GENETICIST			
No	70 (80,45%)		
Yes	16 (18,4%)	RELATIONSHIP	
		Son	5 (31,25%)
		Cousin	4 (25%)
		Father/Mother	3 (18,75%)
		Brother/Sister	2 (12,5%)
		NS	2 (12,5%)
NS	1 (1,15%)		
Total	87 (100%)		
OCCUPATION			
Doctor	43 (49,4%)	PRACTICE AREA	
		Genetics	12 (27,9%)
		Pediatrics	11 (25,6%)
		Neurology	7 (16,3%)
		Gynecology and Obstetrics	3 (7%)
		Family and Community Medicine / Oncology	2 of each (9,3%)
		Anesthesiology / Medical Clinic / Coloproctology / Gastroenterology / Orthopedics / Pathology	1 of each (13,9%)
Dentist	8 (9,2%)		
Nurse	4 (4,6%)		
Biologist	3 (3,45%)		
Academic	3 (3,45%)		
Pharmacist/Psychologist/ Physiotherapist / Health Technician	2 of each (9,2%)		

Architect/Biomedical/Nutritionist/Laboratory Technician/Nursing Technician	1 of each (5,7%)		
NS	13 (15%)		
Total	87 (100%)		
CARES FOR A PATIENT WITH A GENETIC DISEASE			
Yes	61 (70%)	ACCOMPANIES WITH A GENETICIST	
		Yes	42 (68,85)
		No	19 (31,15 %)
No	26 (30%)		
Total	87 (100%)		

*NS: Not Specified

Chart 1. Distribution of health professional registrations at TelegeneticaMG in Minas Gerais by macroregions of Minas Gerais.



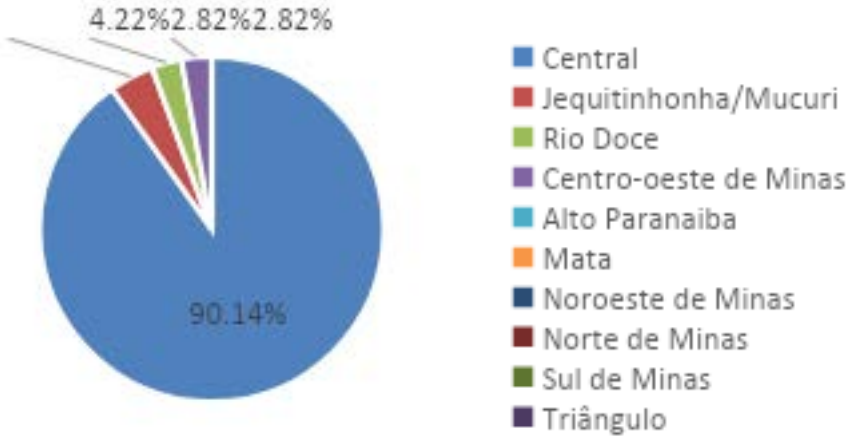
Medium Blue: Central
Light Blue: Triangle
Orange: Central-West of Minas Gerais
Green: Northwest of Minas
Grey: Alto Paranaíba
Yellow: Forest
Brown: South of Minas

Table 2. User profile (patients, relatives and general public) registered on TelegeneticaMG *website*

SEX		
Feminine		67 (75,3%)
Masculine		22 (24,7%)
Total		89 (100%)
STATE WHERE ONE LIVES		
MG		73 (82%)
SP/RJ		4 (4,5%)
RJ		3 (3,4%)
PR/GO/AL/RS/BA/MA/SC		1 of each (7,9%)
NS		2 (2,2%)
Total		89 (100%)
HAS THE DIAGNOSIS OF A GENETIC DISEASE		
Yes	24 (27%)	ACCOMPANIES WITH A GENETICIST
		7 (29%
		17 (71%)
No	62 (69,65%)	
NS	3 (3,37%)	
Total	89 (100%)	

HAS A FAMILY MEMBER WITH A GENETIC DISEASE					
No	37(41,6%)				
Yes	51 (57,3%)	RELATIONSHIP		ACCOMPANIES WITH A GENETICIST	
		Son	27 (52,95%)	Yes	20 (39,2%)
		Mother/Father	8 (15,7%)		
		Brother/Sister	2 (3,9%)	No	26 (51%%)
		Nephew/Niece/ Grandfather/ Grandmother/ Cousin/Father or Mother in Law	1 of each (7,85%)	NS	5 (9,8%)
		NS	10 (19,6%)		
NS	1 (1,1%)				
Total	89 (100%)				
KNOWS SOMEONE THAT HAS A GENETIC DISEASE					
Yes	54 (60,8%)				
No	25 (28%)				
NS	10 (11,2%)				
Total	89 (100%)				

Chart 2. Distribution of the registers of patients, their families and general public in TelegeneticaMG in Minas Gerais by macroregions of Minas Gerais



Medium Blue: Central
Light Blue: Alto Paranaíba
Purple: South of Minas
Orange: Jequitinhonha/Mucuri
Green: Forest
Light Brown: Triangle
Grey: Doce River
Yellow: Central-West of Minas Gerais
Dark Brown: North of Minas

Only 27% of the participants had some type of genetic disease and, of these, 29% had follow-up with a geneticist (Table 2). Most of the participants had relatives with diagnosed genetic diseases (57.3%).

The results show that there is interest from the general population and from doctors, especially pediatricians, in rare diseases of genetic origin. The fact that most of the individuals registered on the website are from the central macro-region of Minas Gerais (which covers Belo Horizonte and the metropolitan region) is explained by the greater dissemination in this area, where reference services in Medical Genetics are located. This fact shows that there is the possibility of growth of the service to other regions of the state.

Only 29% of the patients who claimed to have been diagnosed with genetic diseases were accompanied by geneticists. It is possible to notice the gap between diagnosed patients and Medical Genetics care in the studied population.

Conclusion

TelegeneticaMG appears as an innovative proposal in SUS in Minas Gerais. Many studies point to the need for the use of online education tools in genetics in order to reduce the barriers of lack of specialized professionals, excess of patients in need of care and great territorial distance [5,6,7,8].

The main limitation of the present study is the selection bias, since the participation of the users happened on a voluntary basis. Other evaluations are necessary, aiming additional conclusions.

However, it is possible to affirm that the partial results point to the need of continuous development of telegenetic tools.

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Declaration of conflict of interest
Nothing to declare

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