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ORIGINAL

Training competencies in dysmorphology: learning needs for professionals involved in clinical genetics

Competencias formativas en Dismorfología: necesidades de aprendizaje para profesionales relacionados con la Genética Clínica

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ABSTRACT

Introduction: recent advances in molecular genetics and the use of artificial intelligence require professionals whose work is related to clinical genetics to have a basic knowledge of dysmorphology. It is therefore necessary to design a teaching strategy to improve their professional performance.
Objective: to identify learning needs in dysmorphology for professions related to clinical genetics.
Method: a descriptive cross-sectional observational study was conducted on a non-probabilistic accidental sample of 80 professionals who attended the face-to-face conference "Dysmorphology: current challenges" and who were willing to respond to an anonymous printed questionnaire. Descriptive statistical methods were used to process the data.
Results: the average age of those who responded to the questionnaire was 46,83, the majority were female (92,5 %), predominantly doctors (48,75 %), with an average of 23 years of professional experience. 51,25 % were in teaching positions and 52,5 % were scientists. Most attendees were satisfied with the conference, and the majority of proposals for postgraduate activities to be carried out during the course (63,3 %) included the use of technology for diagnosis (100 %) and the use of nomenclature in dysmorphology (95,5 %).
Conclusion: the handling of nomenclature in semiology and the application of technology for diagnosis were identified as learning needs.

Keywords: Distance Education; Satisfaction; Prevention; Congenital Defect; Phenotype.

RESUMEN

Introducción: los recientes progresos en Genética Molecular, y el uso de la inteligencia artificial, requieren que los profesionales cuya labor sea afín a la Genética Clínica tengan conocimiento sobre fundamentos en Dismorfología, por ello se precisa diseñar una estrategia docente, a fin de incrementar en ellos su desempeño profesional.
Objetivo: identificar las necesidades de aprendizaje en Dismorfología para profesiones cuya labor sea afín a la Genética Clínica.
Método: se realizó un estudio observacional descriptivo transversal en una muestra no probabilística

accidental de 80 profesionales que asistieron a la conferencia presencial "Dismorfología retos actuales",que estuvieron dispuestos a responder a un cuestionario impreso y anónimo. Se emplearon métodos de estadística descriptiva para el procesamiento de los datos.
Resultados: la media de la edad de los que respondieron el cuestionario fue de 46,83, en su mayoría fueron del sexo femenino (92,5 %), predominaron los médicos (48,75 %), y con la media de experiencia profesional de 23 años. El 51,25 % tenían categoría docente y el 52,5 % científica. La mayoría de los asistentes quedaron satisfechos con la conferencia y predominó entre las propuestas de actividades de posgrado a realizar el curso (63,3 %), y que su contenido incluyera fundamentalmente el uso de la tecnología para el diagnóstico (100 %), y el manejo de la nomenclatura en Dismorfología (95,5 %).
Conclusión: se identificó como necesidades de aprendizaje el manejo de la nomenclatura en semiología, así como la aplicación de la tecnología para el diagnóstico.

Palabras clave: Educación a Distancia; Satisfacción; Prevención; Defecto Congénito; Fenotipo.

INTRODUCTION

Dysmorphology is an emerging field of study in Clinical Genetics. It deals with the semiological and nosological analysis of congenital disabilities, in which the collection of information through questioning and physical examination is essential, the latter with special attention to the alterations of disproportionate growth of one region concerning another due to a globally altered morphogenesis, also known as dysmorphic signs.[1,2,3]

Recent advances in Molecular Genetics and the use of artificial intelligence have increased the understanding of the meaning of dysmorphic signs, from which the necessary investigations can be deployed to conclude an accurate diagnosis, allowing the deployment of preventive measures based on Genetic Counselling.[4,5,6]

One of the problems that hinder the application of these technological advances is that the professionals whose work is related to Clinical Genetics and who make up the trans and interdisciplinary study team in the care of patients with congenital disabilities do not have sufficient knowledge about the identification of genetic semiological aspects, and especially the identification of dysmorphic signs; which generates a lack of motivation in their interaction with the Clinical Genetics services, and in the long run an incorrect use of the technology. Therefore, a teaching strategy must be designed to increase their professional performance.[7,8]

As the purpose of postgraduate education is to contribute to the permanent training and systematic updating of university graduates through continuous training and the improvement of their knowledge,[9] this research is carried out to identify the learning needs in Dysmorphology for professions related to Clinical Genetics.

METHOD

A cross-sectional descriptive observational study was carried out on the attendees of the face-to-face conference 'Dysmorphology: current challenges,' which was given by the same speaker at three events:

1. I Mayabeque 2024 Rare Diseases Conference.
2. VIII International Workshop on Rare Diseases Cienfuegos 2024
3. Diabetes 2024

The contents of this conference included:

- Fundamentals of Medical Genetics
- General aspects of Dysmorphology
- Current challenges imposed by technology.

An accidental non-probabilistic sample of 80 professionals registered for these events, who were willing to answer a printed and anonymous questionnaire, previously validated by one of the authors of this research and discreetly modified, according to the particularities of this study, by adding a final open question, was studied.[10]

The instrument consists of 12 questions, 6 of which are closed-ended and one of which is a multiple-choice Likert scale. Among the open questions, one was included regarding the recommendation that would be proposed in new teaching activities on Dysmorphology (see Annex 1). This instrument took into account epidemiological and geographical variables, including gender (male and female), age, and province of residence. All were categorized as nominal qualitative, except for age, which was considered quantitative.

Academic variables were included: scientific degree/academic category (Doctor of Science, Master of Science and those without); profession (nursing, medical, other professions): both polytomous nominal qualitative variables; teaching category (instructor, assistant, assistant, tenured), scientific category (research assistant, research associate, research assistant, and tenured researcher); both categorized as ordinal qualitative variables. Finally, the years of work were considered (0-20, 21-40, over 40 years). The latter was defined as

discrete quantitative.

The level of satisfaction was defined as a Likert-type variable in the following categories: very satisfactory, satisfactory, more acceptable than unsatisfactory, neither satisfactory nor unsatisfactory (or undecided), more inadequate than adequate, and very unsatisfactory.

Additionally, a content analysis was made of the responses to the open-ended question on the recommendation they proposed for a postgraduate activity in this discipline to identify the motivations. These were classified into the type of postgraduate activity proposed and the content in which they were interested in obtaining more information; in all cases, their profession was taken into account.

Data Processing Techniques

The database and its appropriate statistical processing were created using the statistical package Statistical Package of Socials of Science (SPSS), version 29.0.

Statistical methods

Percentage and absolute frequency were used for ordinal qualitative and nominal qualitative variables, and mean and standard deviation were used for quantitative variables.

Ethical Considerations

The research results belong to a project discussed and approved by the Scientific Council and Ethics Committee of the National Centre of Medical Genetics. An informed consent form was drawn up to respect the autonomy of those attending the conferences to participate in the research. Confidentiality of the information obtained when applying the questionnaires carried out throughout the research was maintained.

RESULTS

Of the 80 attendees at the different events who answered the questionnaire, 74 were female (92,5 %). The mean age was 46,83 years, with a standard deviation of 11,74. The mean years of professional work experience was 23, with a standard deviation 12. Of the 80 respondents, 38 (47,5 %) were from Cienfuegos, 28 (35 %) were from Mayabeque, 12 (15 %) were from Havana, and the remaining two were from Matanzas and Holguín (1,3 %, each province). About the professions, 39 were doctors; 10 had a degree in nursing, and the rest had other professions; 42 had attained scientific status (Master of Science 46,3 %, Doctor of Science 3,8 %, and with both 2,5 %). 51,25 % had obtained some teaching category: six were instructors (7,6 %), 20 assistants (25 %), 11 assistant professors (13,75 %), and four full professors (5 %). Of the total, 19 had scientific status, of whom two were aspiring researchers (2,5 %), 12 were Research Associates (15,1 %), two were assistants (2,5 %), and three were tenured (3,8 %).

The conference evaluation by the attendees was satisfactory irrespective of profession, scientific and/or academic degree, and scientific category. However, depending on the profession, the evaluation given by other professionals was in the undecided range (neither satisfactory nor unsatisfactory). The same result was obtained for those in the teaching assistant category.

Responses to suggestions for new teaching activities on Dysmorphology were received from 22 of the 80 respondents, of whom 19 were doctors, one a nurse, and two from other professions.

They made the following proposals regarding the type of postgraduate teaching activity to be carried out: courses: 63,63 %, diploma: 9,09 %, and Master's degree: 27,27 %.

The topics of interest they proposed to address in these activities included the use of technology for diagnosis (100 %), management of nomenclature in Dysmorphology (95,45 %), and Basic Aspects of Genetics (27,27 %).

DISCUSSION

Genetically caused diseases are of low frequency and require diagnosis and management, with the collaboration of professionals from various disciplines who integrate their knowledge in a team effort to achieve a common goal.[11,12]

In this research, the current principles of pedagogy are applied, which include the search for learning needs through individual exploration as the axis of the curricular design of postgraduate training.[13]

Most respondents were female, which corresponded to the composition of health professionals.[14] Age and years of practice reflect that those attending these events are mostly experienced professionals interested in academic exchange. However, there is also a smaller group of young people with a scientific interest.

The three most represented provinces (Mayabeque, Cienfuegos, and Habana) correspond to where the events mentioned above took place, the numbers in each of them being about the number of people attending these events. In turn, most of those who attended were doctors and nursing graduates, corresponding to the majority of university graduates in the National Health System.[14] The proportion of participants with a teaching category and scientific degree was higher than those with a scientific category, as this is a higher qualification.

About the level of satisfaction, the hesitation reflected by other professionals (who were not doctors or nursing graduates) may be related to their lack of motivation derived from their lack of knowledge of fundamental aspects not only of Dysmorphology but even of basic Semiology; however, these same individuals collaborated by answering the last question, which shows that they understood the importance of mastering the practice of physical examination, with the intentional search for dysmorphic signs, and knowledge of the current nomenclature to designate them.

The dysmorphological physical examination is essential to characterize the morphological variants, as a result of which it is necessary to record the precise clinical description, with the use of unified terms to be able to carry out the analysis that leads to the success of the diagnostic process, which is why it is necessary to apply the standardized nomenclature in dysmorphology, included in the HPO (Human Phenotype Ontology), the main ontology that integrates all the phenotypic abnormalities described, which is essential for making decisions regarding the use of technology.[15,16]

The proposal of courses as a professional development activity corresponds to the learning needs perceived by the respondents and are sufficient for the purposes required;[17] the fact that Master's degrees were mentioned, related to postgraduate professional training, is possibly associated with the group of graduates in Cuba of Medical Genetics and Genetic Counselling.[18]

The results presented are partial results corresponding to research in progress. With them, the design of postgraduate activities is imposed, fundamentally of professional improvement corresponding to courses that must start from a version that must be submitted to the criteria of experts.

Limitations of this research

Given the type and scope of the events where the conference was offered and the questionnaire was applied, no representative of all the country's provinces was present.

CONCLUSION

Learning needs were identified for managing nomenclature in Semiology and applying technology for diagnosis.

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CONFLICT OF INTEREST

The authors declare that they have no conflicts of interest.

AUTHOR CONTRIBUTION

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SUPPLEMENTARY MATERIAL 1

We would like your cooperation in filling out this questionnaire in order to know the overall satisfaction rate, strengths and weaknesses in order to take them into account in similar experiences. We appreciate your sincerity and thank you very much for your cooperation.

SEXO	M		F		Age in years:_____	Years of work _____ PROVINCE _____				
Doctor YES ___ NO _____ If other profession, specify: _____										
MASTER OF SCIENCE	YES		NO			Dr IN SCIENCE	YES		NO	
TEACHING CATEGORY	YES		NO			If available, specify: _____				
Research Category	YES		NO			If available, specify:: _____				
HOW SATISFIED ARE YOU WITH THE CONFERENCE?										
Very satisfied										
More satisfied than dissatisfied										
Neutral										
More dissatisfied than satisfied										
Very dissatisfied										
Inconclusive										
Propose any suggestions for new teaching activities on Dysmorphology.:										

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