

SKILLS AND COMPETENCIES IN CLINICAL GENETICS FOR THE EUROPEAN GENERAL PRACTITIONERS

Authors

Dr. Ejarque

Ismael Ejarque Doménech.
Centro de Salud de Almacera, Valencia,
Spain.
Hospital Vithas Aguas Vivas, Alzira,
Valencia, Spain.

Dr. Martín.

Vicente Martín Gutiérrez.
Centro de Salud de San Juan de
Aznalfarache, Sevilla, Spain.

Dr. Cuenca.

Carolina Cuenca Valero.
Centro de Salud de Tres Forques, Valencia,
Spain.

Evidence confirms that diseases are distributed on a spectrum in which genes and environment are closely intertwined. The integration of genetics in primary care requires the gradual incorporation of certain specific elements of clinical genetics, genomics, molecular biology and precision medicine.

Clinical genetics is the implementation of all knowledge and activities in genetics for the benefit of the health of each individual but also of the entire population. It consists of two main activities: 1) clinical assistance for the sick person, at risk of being sick, or of having affected children; 2) focused on the entire population, which is informative-preventive.

In acknowledgement of these limitations, in 2006 it was created the working group semFYC - Spanish Society of Family and Community Medicine - of “clinical genetics and rare diseases”. In 2007, the Group published a paper referring to ten competencies that a Spanish General Practitioner (GP) should carry out for the effective management of clinical genetics related problems. In 2008, a second paper was released about the management of the inherited cancer in primary care, and in 2016 we added new skills in clinical genetics for the Spanish GPs. The Group recognizes that the UK is the only European country with these skills and competencies in clinical genetics in the formative program of their GPs. It is considered essential to incorporate knowledge of clinical genetics into the regular training curriculum. In June 2019, the Commission National de Medicina Familiar y Comunitaria (part of the Spanish Ministry of Health) asked to our working group about these competencies and skills in this matter that should be reached in the formative period of the Spanish GP. From our working group, we are adding efforts to change this situation, aware

that incorporating this knowledge is a commitment to the future for the challenges in the practice of Medicine. Overall, the working states that every European GP should know and apply these skills and competencies to develop correctly their role. The European Society of Human Genetics has also several recommendations in this sense.

I. THE COMPETENCIES ARE

1. Identification of individuals at risk for a genetic condition.
2. Preconception counseling from primary care.
3. Knowledge of prenatal diagnosis techniques.
4. Teratology: to know the phone and the bibliographic resources.
5. Clinical follow-up of the patients with a genetic condition.
6. Identification of psychosocial problems in the rare disease's context.
7. To know the different kind of genetic testing.
8. Properly use the sources of information available at all times.
9. To know the clinical references centers and how to inform the patients.
10. Be aware of one's own limitations.

II. THE SKILLS ARE

1. To know how to draw a detailed three-generation-pedigree according to standardized nomenclature.
2. To understand the inheritance patterns.
3. Continuous updating of the reference centers in clinical genetics.
4. To know the most frequent rare diseases in primary care and to identify the 10-15 rare diseases of each quota – supported by DICE-APER protocol (https://dice-aper.semfyce.es/?page_id=37&lang=en).
5. Proper management to communicate to the patient .
6. To respect the legal and ethical limits regarding the genetic status of a person.

The working group stands for the necessity that a European GP, during his/her training period, should carry out a shift of a month in the reference genetics units.

Out of these competencies, one of which is of greatest interest is counseling or genetic counseling.

An accurate understanding and adaptation to the physiological, medical as well as possible family consequences for a potential genetic condition can be very much provided by genetic counseling. In this sense, communication is also key. As the possible disease may affect several family members with different conditions and level of maturity. Therefore, well trained professionals would definitely help a family under study to:

- Provide a trained analytical review to the family's medical history in a way that potential risks can be assessed, anticipating recurrence for cancer or similar disease types.
- Enrich their understanding of diagnosis parameters such as genetic testing as well as dealing with prevention and, in general, allow proactive research.

- Accompany the patient and its family to make educated decisions along the different stages of the disease, if any.
- Provide with trained support to both the patient and their relatives involved in the process.

A number of core items that are normally developed under the framework of a genetic counseling include, as the first stage, the collection of relevant data from both the patient and the relatives involved. As such, the broad medical history at family level as well as the communication toolkit for accompanying the patient in the process. Also, while it is not necessarily part of the main process, a genetic test might become a possible option. However, it is fundamental that the patient makes an educated decision whether he/she wants to do it or not. That decision has to be built based on trust with the genetic physician assessing benefits and risks that might arise after making the test.

Nowadays, it can be firmly stated that genetic testing as well as molecular diagnosis is growing within the recurrent diagnose and counseling tools of general practitioners. This is in line with other growing trend linked to personalized medicine, where genes play a key role in not only diagnosing, but also preventing present and future diseases.

A notorious faster pace in the development of more accurate genetic tests and tools, as well as the access to larger know-how on genetics and genetic-based diseases, gives society with a very strong tool to diagnose and find the best treatment to diseases that just some years ago were hard to abate. This, accompanied with more mature genetic counseling techniques, might bring a novel tool in form of prevention for certain diseases.

Therefore, the aforementioned advancements in genetics as well as in genomics and proteomics, are making, in general, a great difference with regards to the current advantages to prevent diseases as well as deploying more accurate treatments to patients.