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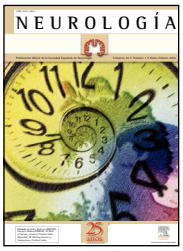

EDITORIAL

Genetic counselling in Neurology: A complex problem that requires regulation

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Abstract
Introduction: The generalization of genetic studies is transforming the practice of Neurology and confronts the clinical departments with new challenges, such as the organization of genetic counselling. The requirement of specialized knowledge, both clinical and molecular, as well as the need for psychological evaluation and family support, especially for predictive testing and reproductive planning, makes a multidisciplinary approach mandatory.
Development: The main characteristics of genetic neurodegenerative diseases are the high level of required specialization –since these disorders are often rare and of difficult diagnosis – together with a generally progressive course, unavailability of effective treatment, the issues generated by predictive testing and the interpretation of genetic testing. The aim of genetic counselling is to provide sufficient and objective information for each individual to make their own decision on genetic testing. It must touch upon psychological aspects and family communication. The PICOGEN program from the Clinic Hospital in Barcelona for genetic testing and counselling of dementias is a good example of integrated strategy capable of managing this new clinical scenario in neurology. Unfortunately, this program is an exception in Spain and the patients with neurogenetic disorders and their families usually do not have guaranteed access to an appropriate care.
Conclusions: Genetic counselling is a unique clinical activity that requires provision of enough time, space and resources to be developed. It implies multidisciplinary participation, due attention to psychological and family issues, and cannot be carried out adequately in a routine Neurology clinic. Legislation is needed to promote a correct articulation of genetic counselling in our country with guarantee of quality and equity.

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This includes training of the necessary health professionals, clarification of competences and provision of resources to the institutions for the development of such programs.

PALABRAS CLAVE
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Enfermedades raras;
Test predictivos

Asesoramiento genético en Neurología: un problema complejo que necesita regulación

Resumen
Introducción: La expansión de los estudios genéticos está transformando la práctica de la Neurología y enfrenta a los servicios clínicos con nuevos retos, como la articulación del asesoramiento genético. La amplitud de los conocimientos tanto clínicos como moleculares precisos, así como la necesidad de una evaluación psicológica y apoyo familiar, especialmente en los análisis predictivos y planificación reproductiva, hacen necesario un enfoque multidisciplinar.
Desarrollo: Las características principales de las enfermedades neurodegenerativas de base genética son el elevado nivel de especialización requerido —por tratarse de enfermedades poco comunes y de difícil diagnóstico— junto con su carácter generalmente progresivo, la ausencia de tratamientos eficaces, la problemática generada por la posibilidad de estudios predictivos y la interpretación de los resultados genéticos. El objetivo del asesoramiento genético es proporcionar la información suficiente y objetiva para que cada individuo pueda tomar sus propias decisiones sobre el estudio genético. Debe incluir la evaluación de aspectos psicológicos y de comunicación familiar. El programa PICCOGEN del Hospital Clínic de Barcelona para el análisis y asesoramiento genético en demencias es un buen ejemplo de una estrategia integrada capaz de abordar esta nueva situación asistencial en Neurología. Lamentablemente, este programa es una excepción en España y los pacientes con enfermedades neurogenéticas y sus familias no tienen garantizada habitualmente una asistencia adecuada.
Conclusiones: El asesoramiento genético es un acto clínico *per se*, que precisa de un espacio, tiempo y recursos suficientes. Implica una participación multidisciplinar, atención a los aspectos psicológicos y familiares y no se puede llevar a cabo correctamente en el seno de una consulta rutinaria estándar de Neurología. Es necesaria una legislación que garantice la adecuada articulación del asesoramiento genético en nuestro país con criterios de calidad y equidad. Esto incluye la regulación de la formación de los profesionales necesarios, la clarificación de competencias y la dotación de recursos a las instituciones para el desarrollo de estos programas.

Introduction

Neurology is one of the specialities in which the progress in health care genetic knowledge is most noticeable as it is transforming our understanding of nervous system disorders. Since the identification of the first genes triggering neurological diseases in the nineteen-eighties, we have lived through an exponential increase in understanding of the molecular bases of a large number of illnesses and genetic alterations giving rise to susceptibility, as well as discoveries on the complexity of our genome and its regulatory mechanisms that are taking us ever further from a simplistic view of the relationship between genotypes and clinical manifestations. To this we have to add the dizzying progress in analytical techniques, with genotyping platforms, sequencing and structural studies with a higher and higher throughput, from which we obtain data requiring expert training for their interpretation. Although the neurological community is still waiting for this knowledge to be translated into effective forms of treatment based on the correction of damaged cell functions, the possibility of completing the diagnosis and prognostic guidance for patients through genetic studies is already a reality neurologists are applying in our daily practice.

The imbalance in the habitual use of genetic tests among neurologists as a group is influenced by numerous factors, including: a) uneven (and sometimes insufficient) training of neurologists in the genetic bases for health and illness; b) variable degree of access to genetic testing in the different regions and centres of our country; c) scant development of clinical trial practice guidelines and recommendations for genetic testing and genetic diseases;

d) lack of adequate regulation for the training in and practice of Clinical Genetics. Regardless of how much it is used and the criteria applied, the availability of genetic testing represents a new fact of life in the medical care of neurological patients: the need for adequate genetic counselling of patients and their families. In the present issue, Fortea et al. review the experience over the last five years of the programme for genetic counselling in dementias at the Clinic Hospital in Barcelona (PICCOGEN).[1] The most outstanding virtues of the PICCOGEN programme are:

1. Its multi-disciplinary character (neurologists, psychiatrists, psychologists, geneticists).
2. Its structured assessment and monitoring protocol, particularly in asymptomatic cases.
3. Co-ordination and selection of cases at the Neurology Department. Its main limitation stems from the administrative context framing the programme, dependent on philanthropic donations in its initial stage and subsequently tied to research projects. The interruption of the programme for three years reflects the weakness of the economic and administrative scaffolding holding it up.

The problems of caring for rare diseases

Most neurogenetic diseases can be classified in the group called "rare diseases": those whose prevalence does not exceed 5 cases per 10,000 inhabitants in the European Union (EU).[2] Despite this low prevalence, the large number of diseases involved means they are reckoned to affect 6% to 8%of the population, around 29 million people in the EU and 3 million in Spain.[3] They are generally chronic, disabling diseases for which the therapeutic resources are generally limited. In recent years, health-care administrations have favoured research into these diseases in order to achieve more effective prevention, diagnosis and treatment. Contributions to this aim have come, for example, from the thematic networks for co-operative medical research (RETICS in their Spanish acronym) into rare diseases, the Institute for Research into Rare Diseases (IIER), the Centre for Biomedical Research in the Rare Diseases Network (CIBERER), the State Reference Centre for the Care of Persons with Rare Diseases (CREER) and the European Networks of Reference Centres for Rare Diseases. Mention should also be made of dissemination actions, databases and registries in connection with this research such as ORPHANET for rare diseases and "orphan" medicinal products (i.e. of little or no commercial interest) as well as the development of quality protocols for genetic diagnosis (for example EuroGentest). All these efforts are aimed at providing a solution for the issues for which patients and their associations demand the greatest attention: the difficulties for obtaining an accurate diagnosis without undue delay, the need for multi-disciplinary attention, access to testing, complex treatment and research, and also on-going information and support. The Spanish health-care strategy for rare diseases as published in 2009 by the Ministry of Health and Social Policies[3] declares the following action lines to be high-priority:

1. Information on rare diseases and available resources.
2. Prevention and early detection.
3. Health-care provision.
4. Therapies.
5. Social and medical attention.
6. Research.
7. Training.

The development of multi-disciplinary units and reference centres capable of covering all care-related aspects of a rare disease or a group of rare diseases is the most suitable and cost-effective way forward. These units must include geneticists and professionals with expertise in genetic counselling.

Predictive studies into late-onset neurodegenerative diseases

Although most genetic diseases debut during childhood or adolescence, with many neurogenetic diseases the clinical manifestations of the molecular dysfunction begin in adult life and follow a progressive course. Genetic tests for these diseases, including the monogenic forms of dementia, are unique in that they can be used for predictive purposes, in other words the result of the genetic analysis can inform us, with a very high degree of probability, whether the person will or will not suffer the disease. It is this capacity to predict serious symptoms without any effective treatment that makes genetic testing so special in Neurology, and obliges us to pay the maximum attention to how these tests are applied and the training of the professionals involved.

Proper attention requires an expert understanding of the clinical situation, differential diagnosis and therapeutic options, but also with regard to human genetics and its biological mechanisms, as well as genetic and biochemical testing, applications and constraints of each technique, genetic databases and search engines as well as the interpretation of the results. Even with strict quality controls, genetic tests are susceptible to methodological or interpretation errors that need to be known.[4] The incorrect interpretation of a genetic result may lead to a mistaken diagnosis, deprivation of specific treatment and even reproductive decisions with catastrophic consequences.[5] Training in genetic, biochemical and molecular biology expertise is thus essential in the team of professionals responsible for the care of these patients. In the same way, it is not possible to imagine adequate attention in line with the predictive study of neurological diseases exclusively by professionals trained in the genetic aspects but in the clinical ones.

Whether by genetic testing or non-genetic tools, the power to predict whether an asymptomatic individual presents a very high risk of suffering a disease has advantages and disadvantages that must be duly discussed, as the person is frequently not aware of all of them. The possible gains from predictive tests include preventive measures or more exhaustive medical follow-up in order to avoid misdiagnoses and unnecessary complementary tests, psychological or social benefits such as better personal and family planning and the benefit that other members of the

family may see their own risk as increased or lowered.[6-8] The problematic aspects of predictive tests include the potential negative psychological impact, but sometimes also the difficulty of interpreting the genetic results. On occasions, variants of unknown significance are observed and it is not possible to be absolutely sure whether or not they are the cause of the illness. In addition, it is impossible to identify accurately the age at onset, the type or severity of the symptoms. Clinical heterogeneity, even within the same family, is frequent in neurogenetic diseases. Complicating even more the genotype-phenotype relationship, the existence of individuals within a family with similar symptoms may be due to coincidence with another cause. This is not unusual in frequent pathologies such as dementia, epilepsy or Parkinson's disease and may significantly hinder the interpretation of the genetic result and the tests' predictive ability. In the specific case of diseases caused by triplets, genetic analyses may detect pathological expansions with a tendency to increase its size even more in the following generation, which might be a cause of "anticipation" of the disease and thus lead to being able to predict a greater risk than in the previous generation, including the risk of serious congenital forms.

The concept of predictive study does not only affect genetic testing

Albeit not always applied, there is considerable consensus about the protocol for assessment, counselling and informed consent before analyzing whether the person at risk is carrying or not the mutation causing the neurodegenerative disease in their family.[9,10] These protocols are aimed at helping to decide whether or not they wish to know their risk status and so provide sufficient information so as not to cross the line of the basic principle of respect for personal freedom, i.e. the right of individuals to take significant decisions about their own lives.[11] Depriving people of the information they need to be able to make a judgement is an attack against their right to freedom. During counselling, they are informed that, as a result of the testing, they can obtain knowledge about their future, and that, once known, there is no going back.

However, the stress is often placed on the predictive capacity of genetic analysis, ignoring the information derived from other complementary tests, structured anamnesis, examination and even simply from a brief meeting with an observant clinical expert. An attentive neurologist receiving a visit from a person at risk for Huntington's disease, fronto-temporal dementia, Charcot-Marie-Tooth syndrome or myotonic dystrophy is able to predict with a high degree of reliability what the outcome of the test will be by observing the attitude, hands, gait, voice quality or a facial expression. Such clinical observation represents in some cases a predictive study as reliable (with similar positive and negative predictive values) as genetic analysis. What should clinicians do with this information?

The ethical and psychological aspects implicated in these situations are complex, especially if patients do not come to the clinic due to their symptoms, in other words for a potential genetic analysis of their case for reasons that have nothing to do with their discomfort. They may not have decided yet whether or not they want to know their risk status. The condition's manifestations are subtle, so neurologists cannot state that a patient is categorically going to present the illness in fact, but only indicate a degree of probability (it should be recalled that genetic testing and any other medical test also indicate a degree of probability, albeit sometimes close to 100%). The dilemma posed is: once the individual has crossed the threshold of the neurologist's office, even if only involuntarily and inevitably, we may have knowledge available about the person (the awareness that he or she is presenting probable incipient manifestations of the condition), knowledge not asked for by the person in question (who has only come for information about the condition of a relative and to assess the options about being analyzed or not); logically, in the case where the patient does come to seek an opinion about symptoms, the dilemma disappears and if no effective preventive treatment can be offered, there would also be no ethical conflict.

Thus, a visit to a neurologist by an asymptomatic person at risk constitutes a potentially predictive "study" yet the patient has not been asked to sign any informed consent form. One could debate whether it is a "predictive" or "symptomatic" situation, as the person is presenting symptoms or signs, albeit very mild or only indicative. Any discussion of these at length should be deferred for another time and place. Let us simply pause to think that, had that person not come into our clinic, he or she would still have some time, perhaps even years, before becoming aware of any illness, even longer until a diagnosis was confirmed and the role of a patient formed part of his or her lifestyle. In the same way as we make sure, or we should make sure, that the individual understands the implications of the information that might be obtained from genetic testing before taking the decision to perform it, would it be necessary to consider a duty of informing them, prior to reaching the neurologist's office, that they are running the risk of obtaining information that they may not necessarily want to receive? What is more, this risk exists even when the examiner says nothing, as a well-executed neurological examination is aimed precisely at bringing to light signs that might otherwise go unnoticed, and they are equally visible to an aware patient at the same time. Similar reflections also apply if we request non-genetic complementary tests for an asymptomatic individual. In the right context, a neurological image or a CPK determination may have practically the same predictive value as genetic testing.

Decision-taking and the role of the psychologist in genetic counselling

In the light of what has been said so far, it can be easily understood why genetic programmes in degenerative diseases have to include the participation of a psychologist. Predictive analyses force individuals at risk to face a complex situation when taking the decision with regard to themselves (should I take the test or not?) and to others (my result changes the risk status of my relatives; who in my

family should I share this information with?, how? and when?). Furthermore, in the case of a young person, delicate reproduction issues might be broached for partners: prenatal analysis, interruption of pregnancy, selection of embryos, sperm or egg donation, adoption, choosing not to have children, etc. Asymptomatic people have many different reasons for considering genetic testing, including: a) uncertainty about their risk generates anxiety, the individual needs to pin down that risk and its implications; b) the desire to plan ahead for vital issues (family ties, employment, relationships[12]). People also often have a strong desire to take control of their lives and participate in actions leading to an improvement in their own expectations and those of others (joining associations, actively seeking information, participating in research and clinical trials).

Psychological support is not limited to carriers. Their partners are the ones left out but they can present as much stress as carriers or even more.[13] In particular, younger generations have difficulty coping with the situation of becoming aware of their genetic status, as this gives them the power and the responsibility to decide about their plans for having a family, they taken on a generational responsibility.[14,15] In a study of 245 individuals who underwent a predictive test for Huntington's disease, the following factors were identified in decisions about having a family: a) the sex of the carrier; b) ethical issues about the prenatal or pre-implantational test; c) the intensity of the desire to have children; d) their prior representation of the disease, as influenced by the experience lived in their own families; and e) the "technological imperative", understood as the feeling of guilt from not making use of techniques available to avoid transmitting the mutation.[16]

The special transcendence of the genetic analyses stems from the fact that the results are not relevant solely for the individual analyzed, but also for other members of the family. For this reason, another fundamental matter is the communication of the genetic information to the rest of the family, as it may have a great impact on the future projects of its members and on their interpersonal relationships. Respecting the principle of confidentiality, the information about the results obtained is usually only notified to the patient who has been studied. Although the 2007 Biomedical Research Act (LIB in its Spanish acronym) reflects that notice should be drawn to the implications the result may have for relatives and the suitability of this information being communicated by the person in question,[17] the decision about whether or not to inform relatives, and who, when and how, remains entirely in the hands of the individual concerned. The information about an index case usually reaches the closest relatives (parents, children, siblings), but not to second-degree relatives.[18] As a result, genetic counselling consultations must include consideration of the subject of communicating the information, helping to identify those family members for whom the information may be more crucial and planning for its notification in an appropriate manner that respects those who do not wish to know. A good understanding of the intra-family dynamics and the factors facilitating and hindering communication may be the key to help them adopt the best strategy for communicating the news to relatives and minimizing the emotional impact.[15]

Genetic counselling: a right of patients and their relatives

The National Society of Genetic Counsellors (NSGC) in the United States put forward the following definition for genetic counselling: "The process for helping people to understand and adapt to the medical, psychological and family implications of the genetic contribution to disease". This process includes the following aspects: a) interpretation of the patient's medical history and that of any relatives to establish the probability of the disease's occurrence or recurrence; b) education about inheritance, analysis, treatment, prevention, resources and research; and c) advice to favour informed decisions and adaptation to risk or illness.[19] Genetic counselling is essential for quality health care in diseases with a genetic contribution, and it must be an integral part of the diagnostic process in neurogenetic diseases. To this end, a specific protocol with multidisciplinary participation is needed since, as has been said, training in various aspects is required, as are appropriate circumstances in terms of facilities and time, matters that are hard for any professional to deal with in isolation.

The fact that genetic counselling process focuses on the family also constitutes a novel element with respect to the traditional doctor-patient (or health-care system-patient) relationship and entails adaptation from a legal and administrative aspect. It is common for different members of a family to belong to different health-care areas. Once the index case has been seen by a particular team, the maximum efficiency is achieved if the rest of the family can be seen at the same centre. This is just common sense: the centre in question has the professionals with the most exact genetic and medical information available about the case for its optimal interpretation; it avoids disparate, or even contradictory, interpretation and information being given to different members of the family; as set out above, the need to understand well the structure and dynamics of the family, with the psychological and social aspects, for the optimal handling of the information and caring for each member.

These quality elements in genetic counselling will be hard to achieve if a family's care is widely dispersed over different centres and professionals. It could even happen that some members of the family might have access to genetic testing and genetic counselling while other relatives belonging to a different centre or region do not.

The 2007 LIB,[17] the law regulating the execution of genetic tests for research and diagnostic purposes, refers to consent, the right to information, the right not to be informed, access to genetic data by health-care personnel, genetic screening and genetic counselling. The LIB indicates that "When a genetic analysis is carried out for health-care purposes, it will be necessary to guarantee that the person concerned receives appropriate genetic counselling, as will be determined in the regulations". It also stipulates that "The whole process for genetic counselling and the practice of genetic analysis for health-care purposes must be performed by qualified personnel and must take place in accredited centres meeting the quality requirements established for the purpose in the regulations". For the first

time, it recognizes the obligation to accompany genetic studies with adequate advice, although it does not establish the mechanisms to ensure its provision. Observing this disposition from the reverse angle, it can be said that the LIB establishes limitations on personal autonomy from the very moment when it only allows genetic analyses to be performed with appropriate advice, restricting individual freedom to proceed with an analysis without the need for genetic counselling. Although the purpose of this restriction is evidently to protect citizens from the possible risk stemming from misinterpreted genetic information and inadequate genetic advice, the option of genetic analyses without obligatorily having to attend a genetic counselling clinic and even without the participation of health-care professionals ("direct to the consumer") is the subject of intense debate. The signatories of this editorial, in particular, are in favour of regulated practice.

The LIB also requires the obtaining of express informed consent in writing for any genetic analysis,[17] thus establishing a clear differentiation between analyses aimed at obtaining genetic information and any other analyses, for which such written consent would not be necessary. The Act clearly includes, however, in this consideration any indirect test allowing the determination of the associated genetic variant (for example, an enzymatic analysis). The fundamental aspects of the informed consent are reflected more generally in the 1997 European Convention on Human Rights and Biomedicine.[20]

It should be said that most of the countries in the EU do not have any regulations specifically governing the genetic counselling process.[21,22] The strictest countries in terms of legislation on genetic counselling include Austria, with a law in force since 1995 declaring non-directive genetic counselling obligatory before and after any genetic test, and requiring it to be carried out by a medical specialist trained in medical genetics. It also establishes that an explanatory report must be delivered to the subject of the study setting out the contents of the genetic counselling sessions.[23] It is noteworthy that the Austrian legislation explicitly protects the right to receive this report, a fundamental document in our opinion, in order to provide the present and future generations of the family and their physicians with very valuable information. The genetic counselling report is as essential a document as it is scarcely ever issued in routine practice, and its goals and contents make it very different from an individual clinical report. In Portugal, for serious diseases debuting in adults with no effective treatment, predictive tests have to be preceded by a psychosocial assessment and follow-up after the result is known.[24] It also establishes that genetic counselling must be provided by medical geneticists and that the government must regulate the supply of genetic tests to avoid the so-called "direct-to-consumer tests", whether by public or private laboratories. The Federal Law on genetic testing in Switzerland sets out in great detail the protocols and applications of genetic counselling.[25] In the United Kingdom, the Association of Genetic Nurses and Counsellors (AGNC) has created an ethical code of conduct with recommendations and "good practices", established the training routes for recognition as a genetic counsellor by the committee created for this purpose and it represents a channel of communication among its associated professionals.

It must also be borne in mind that, in terms of practical applications, Spain is the only EU Member State where the training in Clinical Genetics is not regulated and there is no medical speciality in this discipline. Andalusia was the first region in Spain to regulate the right to genetic counselling through its Law 11 dated November 26th, 2007.[26] This law is intended: a) to regulate the right to genetic counselling, together with the rules developing the same; b) to protect the rights of the persons submitting to genetic analyses, whether for health-care or research purposes; and c) to establish the legal regime governing human DNA banks in Andalusia. Once more, informed consent in writing must be obtained for any genetic analysis, and genetic counselling must be provided, before and after the analysis, to individuals whose samples are to be used for genetic analyses, including a discussion of the repercussions on relatives, therapeutic alternatives and reproductive options. With respect to its application within the scope of the public health system in Andalusia, the Act indicates that:

1. Genetic analyses and genetic counselling must be carried out in the context of comprehensive attention to health care by professionals with adequate skills for the practice of the same on the quality conditions determined by the regulations.
2. The competent department of the Regional Government dealing with health matters will promote training, professional development and qualification of the professionals involved in genetic analysis and counselling, in the framework of the professional development of the Andalusian Public Health System.

It is immediately evident that the development of the LIB, the Andalusian Regional Act and other similar laws implies a need to define what the training of adequate professionals must be like, as well as the practical health-care model to ensure a minimum level of quality in the process. Although the LIB provides a general outline, its development is an urgent necessity.

Need to improve training and qualifications for professionals

One of the great limitations on the development of genetic counselling in our country is the scant genetic content included in the training plans (whether for physician or nurses, for undergraduates or postgraduates) together with the absence of regulated training in Clinical Genetics, a speciality for which the Spanish Human Genetics Association has been fighting for years. Once more Andalusia, within the framework of the Andalusian Genetics Plan, has been the first Region to step up and establish the category of Clinical Geneticist and the procedure to access this newly-created medical category.[27] It provides that Clinical Geneticist may or may not have a degree in medicine and that, if they do have it, they will have the function of providing genetic counselling. This entails the paradox of creating a category of graduates for which there is no

recognized speciality or training programme at the national level. From this Andalusian legislation, it is possible to infer the first positioning of the Spanish administration with respect to who is the qualified professional to undertake genetic counselling, as it stipulates that it must be a graduate in Medicine.

The generalization of this health-care model, then, to a Specialist in Clinical Genetics and not to the other specialists corresponding to the centre with responsibility for genetic counselling. This seems to be the most sensible and realistic approach, at least until such time as the training in Genetics improves in the rest of the specialities. The most recent training programme in Neurology includes the basic concepts of Neurogenetics in the theoretical contents, although it is an optional training spell.[28] Two or three months on a training spell in a Genetics Department are clearly insufficient to acquire the necessary knowledge and skills to cope with reaching a diagnosis and providing genetic counselling. The development of the core subjects and the specific qualification areas within the scope of the Health-Care Professions (Organization) Act ("Ley de Ordenación de Profesiones Sanitarias, LOPS) might be a good opportunity to improve this situation, if Neurogenetics is established as a sub-speciality within Neurology (i.e. neurologists with good additional training in Genetics) and, at the same time, the training in Clinical Genetics and Molecular Genetics is regulated with their corresponding sub-specialization areas (geneticists with good additional training in neurological diseases). These steps would contribute to guaranteeing the existence of professionals with adequate training to offer high-quality health-care programmes to patients with neurogenetic diseases and their relatives and also to ensure the training of the professionals of the future.

Conclusion

Genetic counselling cannot depend on occasional programmes, donations or individual excellence initiatives arising in research groups with their continued extension over time threatened by the funding for their projects, but rather must form an integral part of the routine health care provided to patients and their relatives with neurological diseases of genetic origin and it is inexcusable in particular in predictive and reproductive situations. Our country urgently requires the regulation of all issues referring to the practice of genetics and the training and qualifications of the professionals involved. The Spanish Neurology Society as the duty to give an opinion and participate at all appropriate levels in the decisions and regulations affecting how care is given to patients with hereditary diseases of the nervous system, including the development of the health-care framework for adequate genetic counselling.

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