

eHealth support for genetic diseases: the example of telemedicine

Abstract

To receive medical support from standard clinical services is, in most cases, characterized by inefficiencies and barriers to patients (long waiting times for appointments, time-consumption for the journey to and from the medical site provider, parking, etc.), in particular for some categories of “fragile” people, such as patients affected by a genetic disease, who have to meet their clinicians routinely. The most recent advances in the field of ICT (Information and Communication Technology) are aimed at revising medical practice through new forms of “digital” healthcare, which is expected to improve personalized medical assistance. One of the main strength of personalized assistance is the possibility to offer home-based healthcare. Indeed the digital agenda for Europe 2020 foresees to develop new telemedicine technologies focused to help people affected by genetic and other chronic conditions of living independently even in remote locations. Medical and scientific societies are positively considering innovative approaches, such as the tele-genetics: genetic counsellors and patients will be able to communicate remotely in real time and to share documents (e.g. pedigrees) that are commonly used during in-person sessions.

Keywords: telemedicine, ehealth, genetic disorders, tele-genetics, genetic counselling, digital healthcare

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Abbreviations: WHO, world health organization; ICT, information and communication technology

Introduction

Genetic disorders are chronic and, in most cases, very disabling disorders, characterized by symptoms that persist during the entire life of the affected person. For most genetic diseases, there is no therapy available, and drugs can only bring improvements for some symptoms. According to the WHO, genetic diseases, along with all other chronic diseases, cause at least 86% of deaths in Europe and 77% of health costs (<http://www.who.int>). Moreover, it has been estimated that genetic diseases account for 30-40% of hospitalization in pediatric divisions and are one of the major causes of child mortality in most industrialized countries. Many genetic diseases have an inherited component (transmissible to future generations) and are responsible for a high percentage of morbidity (valued as the impact of a disease within a population) in the adult population, which is affected by a chronic illness on a congenital basis in a ratio of 1:5. For all these reasons, fighting genetic diseases employing innovative eHealth technologies is a top priority of the public health system.

Discussion

Genetic diseases are chronic, and consequently they require a care model substantially different from acute illnesses: local care facilities (namely the “territorial care”) are more efficient than hospitals in the routinely support to patients affected by a DNA defect. In fact, genetic diseases necessarily require a synergic integration hospital-territorial and a multidisciplinary approach between various health professionals and practitioners.¹ Best management should ensure continuity of care irrespective of where the patient is located or how the patient interacts with his/her clinicians. The challenges of this “integrated” care model can be mitigated by the availability of remote information technologies and innovative tools available at relatively low costs. Because of prolonged life expectancy and the increasing number of

people diagnosed with a genetic disease, most health activities should be carried out at home rather than in the hospital. Clinical information and genetic data must be shared quickly between patients and healthcare professionals² as well as home and outpatient clinics. We need to implement technological instruments to measure and transmit clinical parameters even from remote areas and to develop “virtual” visits to reduce the need for frequent hospital admissions that can be very challenging for individuals affected by genetic disorders, who often have also mobility problems. Therefore, tele-monitoring opens new interesting perspectives, with great benefits not only for economic but also for the patients’ quality of life for, which the continuity of care is therefore a top priority. Genetic diseases are also rare, with a prevalence of less than 5 cases per 10,000 people in most of the cases; therefore the patients are often suffering from misunderstanding and isolation of the community in which he/she lives because of the paucity of information and knowledge on genetic diseases and their manifestations. By increasing the digitization processes within the caregiver providers would also facilitate a better awareness of genetic diseases and knowledge of new genomic technologies. Therefore, the Telemedicine, which also includes tele-monitoring, can really help people affected by a genetic defect who need continuous in-home medical health care.

Speeding up the reorganization of mobile assistance by the widespread use of ICT is now imperative. This approach is also important in the digitization of clinical recordings, which is expected to become the unique dataset of all health info and clinical activities related to the state of health of any citizen. The digitizing of clinical information will also improve the sharing of genetic data between researchers and clinicians involved in the fight against genetic diseases; data sharing is commonly believed to be a key tool for advancing therapeutic approaches to these devastating diseases. In the process of digitizing clinical information for genetic data sharing, digital instruments need special attention to protect privacy and personal data. Without the appropriate safeguards for patients, the

potential risks associated with the incorrect use of these innovative technologies could outweigh the benefits. Another fundamental aspect is the need to promote the use of homogeneous technological standards and uniform analysis methods. In addition to the clinical-health aspects, the digitization initiatives will have to be run in parallel with other tools to be developed in the “social” field, which is another fundamental area in the management of people living with a genetic disease.³ Based upon the most recent literature reviews, the benefit of telemedicine programs seems unquestionable, with a 30-35% reduction in mortality and a 15-20% drop in hospitalizations. Reducing recourse to hospitalization, Telemedicine may promote a closer continuity and continuous exchange of information between patient and health care personnel, promoting self-control of the disease and the best compliance with (palliative) treatments. This approach will allow prevention of the worst symptoms and an early recognition of any signs of clinical deterioration. To provide the best healthcare for those who suffer of a genetic disorder requires management skills that will make the already available resources accessible to a broad community of patients and caregivers.⁴ In the field of genetic diseases, the public-private collaboration needs also a better regulation and a closer collaboration. More in general, the digitalization process increases knowledge and skills, enabling system growth and reorganizing the way public health care is conceived. The hospital will also have to change its use of digital technology by taking that technology to the patient’s home.

Conclusion

Telemedicine is a communication system between patient and health care professionals that promotes the continuity of care, allows the intensification of controls, improves the patient’s awareness and the active involvement of his/her family members. The hypothesis is that, for patients suffering of a genetic disease, the prevention of frequent hospitalization, which affects their quality of life, can be obtained through a closer clinical and instrumental follow-up associated with the ability to monitor various clinical parameters easy to be measured.

However, it must be absolutely clear that Telemedicine is additional and not an alternative to the classical medical assistance. Telemedicine should be integrated within a new management model to facilitate, intensify and personalize the monitoring of genetic diseases. To achieve the best clinical benefits, the problem is not the technology, now effective, but the choice of the most suitable patient, clear flow charts and the availability of well-trained personnel with rapid and appropriate intervention skills. Finally, a central issue for the spread of Telemedicine remains the need for an adequate remuneration policy for long-distance reimbursed services.

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Conflict of interest

The authors hold stock of Net-Medicare SrL, an Italian start-up company providing telemedicine services and declare that the ownership does not have any influence on the subject of the present article.

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