

# Health policy and legal regulations concerning the functioning in society of individuals burdened with Huntington's disease and other rare diseases

Daniel Zielonka, Jerzy T. Marcinkowski, Aneta J. Klimberg

*Chair of Social Medicine, Poznan University of Medical Sciences, Poland*

Zielonka D, Marcinkowski JT, Klimberg AJ. Health policy and legal regulations concerning the functioning in society of individuals burdened with Huntington's disease and other rare diseases. *J Pre-Clin Clin Res.* 2013; 7(1): 27–31.

## Abstract

In the European Union, 5,000–8,000 distinct rare diseases affect up to 6% of the total EU population; therefore, care of these patients and the possibilities to optimize this care become increasingly more clearly perceived in health policy. Among these rare and ultra-rare diseases, Huntington's disease (HD) deserves attention in the context of exercising health care over patients suffering from this disease, access to medications, rehabilitation, etc. Legal regulations are being developed concerning the functioning in society of individuals burdened with rare and ultra-rare diseases – beginning at the EU level. Health education pertaining to this scope of problems is necessary, as well as providing society with information concerning health and social problems encountered by patients with rare diseases. In this context, the mass media play an important role. Systemic changes are desirable, including the regulations, and for this reason, a decisive legislative initiative originating from, e.g. associations of patients suffering from rare diseases, may lead to the changes in regulations to the benefit of patients with such diseases, including HD. Unfortunately, to-date, the legislator has not attached much importance to this problem.

## Key words

Huntington's disease, rare diseases, health policy, legal standards, social functioning

Before 1993, individuals burdened with the mutation conditioning the occurrence of Huntington's disease (HD) [1], were frequently unaware of the genetic defect with which they were affected until they experienced the first symptoms of the disease. In consequence, they could not consider in their life plans the disease, which usually manifests itself at middle age, i.e. at the age of 30–50. Their life plans concerning education, selection of an occupation, undertaking of work, payment of contributions to the National Health Insurance Agency (NFZ), did not cover, e.g. the necessity to work for the period of time which would authorize them to use health benefit. As a result, individuals suffering from HD were forced to resign from work and remained without the right to any health allowance [2].

It is estimated, that from the moment of making the diagnosis of HD, the mean survival time is 15–20 years (juvenile Huntington's disease has its onset before the age of 20, and is also characterized by a survival time shorter than average). The frequency of occurrence of HD is estimated to be 4–8 per 100,000 [3, 4].

The sole process of treatment is a great challenge for the health care of patients diagnosed with HD.

Firstly, at present, there is a lack of possibilities of causative treatment of the disease, and the studies to-date have not confirmed the effectiveness of the proposed methods of neuroprotective treatment, which would slow down the progression of HD.

Secondly, access to the most modern drugs for the symptomatic treatment of HD is significantly limited by

the fact that the disease has not been included in the list of chronic diseases, and is only considered as a rare disease [5]. In consequence, while completing a prescription, a physician cannot make a note that the patient is ill with a chronic disease, which, in turn, results in the lack of preference prices for patients with HD (only patients ill with polyketonuria are treated differently) [6, 7, 8].

Thirdly, in the system of health care in Poland, medical assistance and rehabilitation have not been provided on the level which would ensure an efficient control of the effects of the disease. Patients with HD possess the same access to rehabilitation courses, spa treatment, and logopaedic exercises; however, in their case this is definitely not enough. Cases have been noted in which patients with the symptoms of HD were simply refused the referral for additional rehabilitation procedures and exercises, by stating that according to the advancements of medicine the regaining or improvement of health would not occur. In Poland, the maintenance of efficacy and independence of patients for the longest time possible, which is a priority in other European countries, remains a secondary issue [9].

According to the World Health Organization (WHO), based on the WHO Declaration of Patients' Rights and the Charter of Patient's Rights: 'Every individual has the right of access to health services on the level provided by actions in the area of prevention and health care, and use the possibilities to enjoy the highest attainable health level.' In 1994, the WHO presented the Model of Declaration of Patients' Rights as guidelines to be applied in individual member countries [8].

The subsequent international organization with global reach – the United Nations (UN) – in the Charter of Human Rights does not differentiate the characteristics of patients

Address for correspondence: Jerzy T. Marcinkowski, Chair of Social Medicine, Poznan University of Medical Sciences, Rokietnicka 5C, 60-806 Poznan, Poland  
E-mail: jtmrcin@gmail.com

Received: 20 October 2012; accepted: 19 March 2013

(age, nationality, religious beliefs, nosologic unit diagnosed), which would in any way allow the limitation of access to treatment or to the latest achievements in medicine [10].

The European Parliament also considered rare diseases in its regulations. In Regulation (EC) No. 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products, stated as follows:

The European Parliament and the Council of the European Union, having regard to the Treaty establishing the European Committee, and in particular Article 95 thereof, having regard to the proposal from the Commission, having regard to the opinion of the Economic and Social Committee, acting in accordance with the procedure laid down in Article 251 of the Treaty, states as follows: 'patients suffering from rare conditions should be entitled to the same quality of treatment as other patients; it is therefore necessary to stimulate the research (...)' [11].

The European Commission (EC), considering the importance of the problem, also started to deal with the scope of problems concerning rare diseases. In consequence, the European Commission Directorate General for Health and Consumers developed a document: 'Rare Diseases: Europe's Challenges'. A Community action programme on rare diseases, including genetic diseases, was adopted for the period 1 January 1999 – 31 December 2003. This programme defined the prevalence to be low, if a disease affects less than 5 per 10,000 persons in the European Union. This translates into approximately 246,000 persons per disease in the EU with 27 member states [12].

Based on the present scientific knowledge, it is known that 5,000–8,000 distinct rare diseases affect up to 6% of the total EU population at one point of life. This means that approximately 15 million EU citizens, in 27 member states at the time of data compilation, are or will be affected by a rare disease (...):

An individual suffering from a rare disease has the same right to the necessary treatments and medication as someone with a more common disease. The development of a European collaboration for the delivery of health care and medical services to RD patients will have a major potential for bringing benefits to European citizens [12].

The European Charter of Patients' Rights contains 14 patient rights, which jointly lead to the guaranteeing of a high level of human health protection (Article 35, The European Charter of Patients' Rights, Basis Document) and the provision of high quality services provided by various systems of health care in Europe [12].

Within the context of the problem of the right of access to medical care – irrespective of the disease diagnosed in a patient – the following extract from the above-mentioned Charter may be quoted:

Every individual has the right of access to the health services that his or her health needs require. The health services must guarantee equal access to everyone, without discriminating on the basis of financial resources, place of residence, kind of illness or time of access to services [12].

The viewpoint adopted by Decision No. 1295/1999 EC of the European Parliament and of the Council of 29 April 1999, adopting a programme of Community action on rare diseases within the framework for action in a field of public health (1999 to 2003), Clause 13 states the following task:

Whereas rare diseases have been identified as a priority area for Community action in the Commission's communication of 24 November 1999 in the framework for action in the field of public health [13, 14].

Within the European Project for Rare Diseases National Plans, the representatives of 27 European states, including Poland, agreed to elaborate and implement before the end of 2013 the national plans or strategies for tackling the difficult situation of patients suffering from rare diseases. In this document, it is stated that the difficult situation of patients results primarily from the specificity of this group of diseases – some of them occur so rarely that the adequate medicinal products have not yet been developed, and the low level of knowledge concerning these diseases does not allow the making of a correct diagnosis of the disease. Even if in some cases only temporary aid may be provided, in the EU countries patients afflicted with rare diseases are entitled to specialist medical care and rehabilitation. Prior to the implementation of these regulations, the Germans had won financial resources for terminally ill children at the European Court of Human Rights.

EUROPLAN (European Project for Rare Diseases National Plans Development) coordinated by the National Office of Rare Diseases Research at the Italian National Institute of Health, has a goal to facilitate for the Community member states the development of national strategies in the field of rare diseases, aimed at:

- the provision of coherence of national plans and strategies and make such initiatives consistent with a common strategy at the European level;
- monitoring of progress in the implementation of national plans and strategies, and the comparison of data among Member States;

According to the recommendations by the European Commission, the plans should be implemented before the end of 2013. The EUROPLAN, which is being performed within the first project of the Programme of Community action in the field of health, began in 2008 and is planned to be completed in 2011.

According to the monitoring, seemingly the smallest problem concerning access to therapy and resources for rehabilitation was noted in the USA and Japan by means of various projects:

- experimental;
- sponsored;
- clinical (university centres).

Despite this, however, some USA citizens sceptically approached the problem of the effect of the State on treatment and its availability. Therefore, it is worth bearing in mind the quotation attributed to Mark Twain: 'No man's life, liberty, or property are safe while the legislature is in session'.

In Poland, the effect on the availability of health services is in the hands of Parliament, the government, and especially the Minister of Health. The Act in the Matter of Patients'

Rights and Patient Rights Ombudsman of 6 November 2008 is in effect [15]. The problem of rare diseases concerns several thousand patients who have been waiting for years for the implementation of adequate legal regulations, which would enable them to have access to specialist medical care and modern therapies. It is expected that similar to other EU countries, the situation in Poland will be improved by the EUROPLAN project begun in 2008. The national plan of actions in the field of rare diseases was supposed to be designed by 2011 [16]. It is being developed by the Group for the Matters of Rare Diseases created by the Minister of Health. The project would cover the financing of the treatment of rare diseases and increasing the level of care of patients suffering from these diseases. An individual affected by a rare disease, including Huntington disease, often requires round-the-clock care, which is usually associated with the fact that the affected person has to resign from work. Caregivers who exercise care of the patients will be included in the welfare system and receive additional means.

The Minister of Health of the Republic of Poland stated that from April 2011 the Department of Health has been co-financing the international project 'Orphanet', established by the French Ministry of Health [14]. The goal of the project is to increase the access of patients and physicians to information concerning rare diseases. A relevant, already existing and comprehensive database will be translated into Polish. The Minister of Health of the Republic of Poland made the following announced:

1. We will reimburse the subsequent medicines applied in rare diseases' (Interview for TVP Info);
2. We have found 200 million PLN for the treatment of rare and ultra-rare diseases [17];
3. The agreement will be performed with associations of patients with rare diseases concerning a systematic inclusion of subsequent rare diseases in the list of reimbursed drugs [17].

While designing any projects, the following should be considered:

- patient demands (drugs, specialist care, auxiliary aids, rehabilitation courses);
- differentiation of demands according to the stage of the disease (early, intermediate, advanced);
- necessity to support families of patients suffering from rare and ultra-rare diseases;
- need for expanding health education in the area of the fundamental knowledge concerning rare diseases and the possibility to perform tests, e.g. genetic, for the purpose of early diagnostics.

According to estimations, in Poland more than 7,000 patients suffer from so-called common diseases, whereas 750–7,000 – from rare diseases, less than 750 patients. The accessibility to treatment, diagnostics, reimbursement of the costs of treatment and drugs is inversely proportional to the frequency of occurrence of the disease in patients. This situation is due solely to economic reasons. According to Marek Kokot:

A modern physician no longer has the right to approach medicine from the aspects (...) I do not care by what means I struggle for the life and health of a patient

(...) Each medical decision is trading off – honestly speaking, by rescuing an X, I reduce the chances of the Y for treatment (which does not mean to cure) – longer pretending, escape from this important dilemma is simply burying one's heads in the sand [18].

In the face of such an attitude, Polish physicians have many dilemmas concerning:

- number of patients admitted;
- number and type of diagnostic tests performed;
- treatment of patients burdened with rare diseases;
- prescribing rehabilitation (time, type, place).

Increasingly more frequently, due to the Legislator, the concepts a 'patient', 'the ill' disappear from the subsequent Acts, and are substituted by the notion 'care recipient', and consequently – a physician is a 'care provider'.

There occur very difficult questions in the field of ethics, e.g.: How to approach the problem of financing health services, the costs of assisting those who are most seriously ill, the treatment of whom is most often the most expensive? Solidarity, utilitarianism, egalitarianism...? According to Zbigniew Szawarski:

If we adopt a utilitarian theory, all drugs for rare diseases (so called orphan drugs) 'go overboard'. The economic account leaves no place for sympathy.

Egalitarianism is not a much better solution: If everyone has an equal right to health care, this means that everyone has a right to some minimum. The question is: who, in what area, and considering what values establishes this minimum, and if rare diseases will be 'inserted' into this minimum.

The ethics of solidarity also do not provide us with possibilities to select procedures of making moral decisions. One cannot be in solidarity with all of them. A hierarchy of importance must be constructed (...) There will always be groups competing for our solidarity and it will be necessary to make a choice about who should dispense a limited pool of resources. Granting it to one group means taking it from others.

At present, no moral system provides a solution which would be in accordance with our sense of justice. Whatever we choose, we will be haunted by a moral hangover... [19].

The situation of patients with chronic, oncologic, rare disease, including HD, and ultra-rare diseases, is being slightly improved by the State Fund for Rehabilitation of the Disabled (PFRON), non-government organizations, foundations, and anonymous donors.

At the time of advancement in technology there open routes for seeking assistance, informing society about problems concerning certain population groups in some way 'stigmatised' by the disease. This kind of health education, which leads to making society aware that just behind the wall, there may live someone who is ill, disabled, who sometimes needs us to go shopping, post a letter, or sometimes our interest, conversation and understanding and, obviously, also a smile and a good word – they also cure.

The patients' significant others often reach for various methods of making their way to society. These are leaflets distributed in public places, press advertisements,

announcements on various websites, or announcements on the radio. However; so-called intervention programmes in the mass media reach the largest number of people, such as “A Matter for the Reporter”, “Reporters’ Express”, “Attention”, or “Intervention”. Sometimes the participation in such a programme, for a patient and those who are close to him, is the last resort. Is this so because they are not noticed in their environment by medical staff and officials?

Should we strive for changes? In what direction should these changes go? Certainly efforts should be undertaken in the field of the scope of knowledge of society concerning rare diseases, their essence, and the problems of individuals burdened with these diseases. People want to know more about such diseases, their causes, course, prognoses, as well as the life conditions of patients with the diagnosis from individual nosologic units.

While dealing with the scope of problems pertaining to HD (and other genetic diseases), the *in vitro* method should be mentioned, offered to, among others, potential parents in whose families there occur genetic diseases. It seems that this method, although controversial from the point of view of the Church – as a non-natural way of fertilization, and from ethical-moral aspects – as an introduction to eugenics promoted by totalitarian regimes of the 20<sup>th</sup> century, may be used to eliminate, prior to implantation in the uterus, of the cells burdened with mutation which causes the disease. It is necessary to make the healthy and the ill aware that the *in vitro* method is not the only method of infertility treatment, but may also be used for the elimination of risk of transmitting genetic diseases to the offspring. While applying pre-implantation diagnostics it is possible to diagnose single gene and chromosome translocation disorders. People who fear the risk of occurrence of genetic diseases – including HD – increasingly more frequently decide to perform genetic tests. Due to the ethical-moral issues, the costs of these tests are not reimbursed by the National Health Insurance Agency; nevertheless, taking into account an economic calculation, such a reimbursement would, in effect, be considerably lower than the potential reimbursement amounts from the National Health Insurance Agency, nursing allowances, endowments and disability benefit in the case of the occurrence in a child of a rare genetic disease, and the need for many-year subsidizing of such patients and their families.

At present, the phantom of the application of gene therapy in HD is increasingly more clearly evident on the horizon. It has been confirmed that the intentional ‘attenuation’ of the mutant *HTT* gene responsible for the occurrence of the symptoms of HD is a very effective method of treatment in the HD mice model [20]. At the moment of occurrence of the possibilities of application of treatment with the use of gene therapy in humans, the tendency towards systemic changes, alterations in regulations will become important. This would enable the reimbursement for such therapy from the National Health Insurance Agency in the case of patients with the diagnosis of HD, and individuals with mutation confirmed, who have not yet developed the symptoms.

Only a decisive legislative initiative – coming from, e.g. associations of patients suffering from rare diseases, may lead to the change of regulations for the benefit of patients with rare diseases, including HD.

Until now, the Legislator has not attached much attention to this problem. Since 10 February 1919, i.e. the first meeting of the Parliament of the Republic of Poland, not a single

Act has been passed, no directive, regulation, or annex published concerning patients with HD. Since 10 February 1919, neither the Constitutional Tribunal nor the Supreme Court of Justice have passed any resolution in the matter of patients ill with HD.

The following general regulations concern patients suffering from HD:

- granting of health benefits, nursing allowances from the Social Insurance Fund (ZUS);
- decision-making concerning disability;
- support from municipal, communal welfare centres (MOPS, GOPS) and provincial offices for family support (PCPR).

Apart from changes in the regulations, more intense educational activity is necessary concerning the demand for the organization of rehabilitation, logopaedic, dietetic exercises, spreading a ‘protective parachute’ over the families of the patients, organization of support groups for families, in which Huntington’s disease occurred.

## REFERENCES

1. <http://www.huntington.pl/choroba/choroba-huntingtona> (access: 2011.03.21).
2. Ustawa z dnia 1 lipca 2005 r. o zmianie ustawy o emeryturach i rentach z FUS. Dz. U. nr 169, poz. 1412. (Act of 1 July 2005 in the Matter of Change of the Act Concerning Retirement and Health Benefits from Social Insurance Fund (FUS. Official Journal No. 169, Clause 1412).
3. Roos RA. Huntington’s disease: a clinical review. *Orphanet J Rare Dis.* 2010; 20; 5(1): 40.
4. Pekmezovic T, Svetel M, Maric J, et al. Survival of Huntington’s disease patients in Serbia: longer survival in female patients. *Eur J Epidemiol.* 2007; 22(8): 523–526.
5. Lista chorób przewlekłych – zgodnie z rozporządzeniem z 2009 roku. Dz. U. z 2009 r., nr 212 poz. 1647. (List of Chronic Diseases – according to the regulation of 2009. Official Journal of 2002, No. 212, Clause 1647).
6. Rozporządzenie Ministra Zdrowia z dnia 23 grudnia 2010 r. w sprawie wykazu cen urzędowych hurtowych i detalicznych produktów leczniczych i wyrobów medycznych. Dz.U.10.251.1689 z dn. 29 grudnia 2010 r. (Regulation by the Minister of Health of 23 December 2010 in the matter of the list of official, wholesale and retail prices of therapeutic and medicinal products. Official Journal 10.251.1689 of 29 December 2010).
7. Rozporządzenie Ministra Zdrowia z dnia 22 grudnia 2010 r. w sprawie wykazu leków podstawowych i uzupełniających oraz wysokości odpłatności za leki uzupełniające. Dz.U.10.251.1686 z dnia 29 grudnia 2010 r. (Regulation by the Minister of Health of 23 December 2010 in the matter of the list of basic and supplementary drugs. Official Journal 10.251.1686 of 29 December 2010).
8. Rozporządzenie Ministra Zdrowia z dnia 22 grudnia 2010 r. w sprawie wykazu chorób oraz wykazu leków i wyrobów medycznych, które ze względu na te choroby są przepisywane bezpłatnie, za opłatą ryczałtową lub za częściową odpłatnością. Dz.U.10.253.1699 z dnia 29 grudnia 2010 r. (Regulation by the Minister of Health of 22 December 2010 in the matter of the list of diseases and list of drugs and medicinal products which due to these disorders are prescribed free, for lump-sum charge or a partial payment. Official Journal 10.253.1699 of 29 December 2010).
9. Prawa pacjenta i zadośćuczynienie pieniężne za ich naruszenie w prawie medycznym i cywilnym (Patient rights and financial compensation in medical and civil law) <http://www.prawoimedycyna.pl/?str=artykul&id=45&PHPSESSID=00e95a4e4f600de896a5b2ba2d520f39> (access: 2011.04.02).
10. Powszechna Deklaracja Praw Człowieka. (Joint Declaration on Patients’ Rights) [http://www.unesco.pl/fileadmin/user\\_upload/pdf/Powszechna\\_Deklaracja\\_Praw\\_Czlowieka.pdf](http://www.unesco.pl/fileadmin/user_upload/pdf/Powszechna_Deklaracja_Praw_Czlowieka.pdf) (access: 2011.04.02).
11. European Commission. [ec.europa.eu](http://ec.europa.eu) (access: 2011.04.02).
12. Europejska Karta Praw Pacjentów. Instytut Praw Pacjenta i Edukacji Zdrowotnej. (European Charter of Patients’ Rights). <http://www.prawapacjenta.eu/index.php?pid=443> (access: 2011.04.02).

13. Decision No 1295/1999 EC of the European Parliament and of the Council of 29 April 1999 adopting a programme of Community action on rare diseases within the frame-work for action in a field of public health (1999 to 2003). Official Journal of European Communities 22.6.1999 L155/1–5. [http://www.rzadkiechoroby.pl/dodatki/UE%20Decision%201295\\_1999%20on%20rare.pdf](http://www.rzadkiechoroby.pl/dodatki/UE%20Decision%201295_1999%20on%20rare.pdf) (access: 2011.04.02).
14. Choroby rzadkie. (Rare diseases) <http://www.rzadkiechoroby.pl/rzadkiechoroby.htm> (access: 2011.03.30).
15. Ustawa z dnia 6 listopada 2008 r. o prawach pacjenta i Rzeczniku Praw Pacjenta. Dz. U. Nr 52 poz. 17 z 2009 z dnia 31 marca 2009 r. (Act of 6 November 2008 in the matter of Patients' Rights and Ombudsman of Patients' Rights. Official Journal No. 52, Clause 17 of 31 march 2009)
16. TVP Info/rynekapteki.pl (access: 2011.03.30).
17. <http://www.orpha.net/consor/cgi-bin/index.php> (access: 2011.03.30).
18. Analizy ekonomiczne w służbie zdrowia. (Economic analyses in health services). [www.mckp.cm-uj.krakow.pl/szkolenia2010/.../analizy\\_ekonomiczne1.pdf](http://www.mckp.cm-uj.krakow.pl/szkolenia2010/.../analizy_ekonomiczne1.pdf) (access: 2011.04.02).
19. Jakubiak L. Konferencja „Ocena technologii medycznych (HTA) a kluczowe decyzje w ochronie zdrowia”, (Evaluation of medical technologies (HTA) and key decisions in health services) Warszawa, 22 March 2011 r.; Luiza Jakubiak/rynekapteki.pl (access: 2011.04.02).
20. Grondin R, Kaytor MD, Ai Y, Nelson PT, Thakker DR, Heisel J, Weatherspoon MR, Blum JL, Burright EN, Zhang Z, Kaemmerer WF. Six-month partial suppression of Huntington is well tolerated in the adult rhesus striatum. *Brain*. 2012 Jan 16.