

**PRACTICAL EXPERIENCE WITH PROVIDING SPECIALIZED  
SOCIAL SERVICES FOR RARE DISEASE PATIENTS***Madunová A., Duračinská M.***Muscular dystrophy patient organization in the SR**

Organization of Muscular Dystrophy in the Slovak Republic (OMD in SR) is the only specific organization in Slovakia which associates children and adults with muscular dystrophy and other types of neuromuscular diseases (hereafter only NMD) and their families. The organization was founded as an independent public association in 1993. It has been providing social counselling and other specialized social services for its members and clients. In its early years, it performed the community service and non-profit activities on a voluntary basis; today, a professional team of nine takes part in its activities, seven of which are people with extensive disability, plus two assistants. This change could take place thanks to having obtained the status of a protected workplace in 2009. In accordance with EURORDIS recommendations, OMD in SR takes care of activities in 3 different areas: information services and help line, therapeutic recreational programmes, and respite care services.

As a part of the information service, it provides specialized social counselling for disabled people via phone, email, in person or in the field – at a patient's place. The organization also publishes its own quality magazine *Ozvena (ECHO)*, which is distributed free-of-charge to all members four times a year. People with NMD are involved in the creation of the magazine from the position of experts and patients.

The organization is particularly devoted to Recreational activities the camps for children with NMD, especially boys with Duchenne muscular dystrophy. Activities organized every year include also integrative national meetings of members.

The group of relief services includes providing specialized social services in the *Personal Assistance Services Agency* founded under OMD in SR. It provides wide-range service for 180 personal assistance users in Bratislava region; however, counselling covers the whole Slovakia.

For a long time, the organization has been making effort to change regulations in the legislation which concern several aspects essential for people with NMD.

Slovakia is not doing enough to provide financial support for functioning of patient organizations – the subsidy system does not take them into account and donation

support is non-existent. A tool for change could and should be the National Plan for Rare Disease, which will not be really implemented without providing subsidies for patient organizations working in the area of rare diseases. In 2012, the organization became a member of EURORDIS and Alliance TREAT – NMD.

**Keywords:** *neuromuscular diseases – specialized services- social counselling – relief services*

Muscular dystrophy patient organization in the SR (MDPO SR) is the only specialized organization in Slovakia which associates children and adults with muscular dystrophy and other types of neuromuscular diseases (hereafter only NMD) and their families.

The organization was founded as an independent public association in 1993. It has been providing social counselling and other specialized social services for its members and clients. In its early years, it performed the community service and non-profit activities on a voluntary basis. Today, a professional team of nine takes part in its activities, seven of which are disabled people, plus two assistants. This change could take place thanks to having obtained the status of a protected workplace in 2009, which enabled the creation and financing of qualified positions for wheelchair people with muscular diseases by means of active employment support tools. These positions include for example a social counsellor, a project coordinator, and a web administrator (<http://www.omdv.sr.sk/index.php/chranene-pracovisko/zamestnavame.html>).

The organization observes the philosophy that people suffering from NSDs can understand other NMD patients' problems best; and therefore, the executive, managing and control bodies are staffed with people with NMDs or parents of children with NMDs. At the same time, people with NSD are also initiators and executors of individual expert activities and other activities of the organization.

Our organization differs from other patient organizations in terms of promoting the philosophy of independent life into practice, which means equal inclusion of disabled people into all areas of social life and a positive example of coping with the consequences of such disability in relation to the public. These attributes are fulfilled by providing social services, individual activities, and by its philosophy of employing people with NMD which supports activation and full-fledged life of people with dystrophy and other most severe forms of disability.

The quest of the organization is to defend and enforce rights and needs of people with NMD. In order to do so, it actively comments on legislation, also beyond social and health areas. Its main goal is to provide directed and systematic help to the members, which reacts to their gradually deteriorating health. The progress of the disease manifests mainly in the need of physical help of greater extent, necessary barrier-free home modifications, dependency on several types of compensation aids – in case of dystrophy patient with severe lower and upper limbs disability it is for example a mechanical and an electric wheelchair, an adjustable bed with an anti-decubitus mattress, lifting devices, car modifications, such as a platform or a revolving passenger seat, etc.

In accordance with EURORDIS recommendations, the muscular dystrophy patient organization in the SR takes care of activities in 3 different areas: information services

and help line, therapeutic recreational programmes, and respite care services. As a part of the information service, it provides specialized social counselling for disabled people via phone, email, in person or in the field – at a patient's place.

The organization also publishes its own quality magazine *Ozvena*, which is distributed free-of-charge to all members four times a year. People with NMD are involved in the creation of the magazine from the position of experts and patients. For many members, it is still the only source of information on the disease and the related problems (<http://www.omdvsk.sk/index.php/casopis-ozvena/rocnik-2012.html>).

If necessary, the organization will arrange a contact to NMD specialists. The organization has been cooperating with the scientific community for a long time and it has been active in the area of NMD health care and research, as the state does not cater for this area sufficiently.

In 2010 it financially supported the foundation of a joint database of Czech and Slovak Duchenne/Becker muscular dystrophy patients (hereafter DMD/BMD) and the carriers of this disease gene. Creation of the database in the Czech Republic was promoted by the public association Parent Project, which has been managing the database in cooperation with the Institute of Biostatistics and Analyses at Masaryk University in Brno. The database is called REaDy – REgister of muscular dystrophies (<http://ready.registry.cz/>). Currently, the database registers a total of 54 patients from Slovakia, one of which is a woman. The organization then financially supported training of four Slovak paediatric neurologists. Thus, the patients are registered to the database primarily through expert workplaces in Slovakia, which include: MUDr. Roman Mego, Sinalgis, Bratislava; MUDr. Karin Viestová, CHFH Bratislava; MUDr. Katarína Okáľová, CHFH Banská Bystrica; MUDr. Eva Lazarová, CHFH Košice. A part of Slovak patients are registered through FN Brno (15 patients out of the total number). Thanks to the registration, two Slovak boys with DMD were included in the third phase of a randomized study. The clinical study started in December 2010 and the time of completion is estimated to be July 2013. It takes place at 47 locations all over the world; the Slovak boys are studied in Brno. The drug which is being tested is from the group of antisense oligonucleotides and it is administered to patients once a week over the period of one year (<http://clinicaltrials.gov/ct2/show/NCT01254019?term=duchenne+muscular+dystrophy&rank=43>).

MDO in the SR publishes informational brochures, which bring information from the area of health care and rehabilitation for people with NMD and their families in a comprehensive way. (Maduna, M.: Physiotherapy at Home: Shortened Muscles Stretching in Children with Muscular Dystrophy, 2008; Maduna, M.: Physiotherapy at Home: Breathing Rehabilitation in Neuromuscular Diseases, 2010; <http://www.omdvsk.sk/index.php/kniznica/vlastne-publikacie.html>).

In the previous year, it provided a translation of and obtained funds by means of a grant for a publication which brings the standards of health care for DMD patients in Slovak language in a form of a brochure for families. The recommendations involve interdisciplinary care and its management. 84 international experts from different areas of expertise participated in the creation of the document. The publication entitled *Duchenne Muscular Dystrophy: Diagnosis and Treatment* was originally published in English as a part of a European project called TREAT – NMD. The Slovak version

of the brochure is published at <http://www.treat-nmd.eu/dmd/care/family-guide/translations/> and available for downloading.

Muscular dystrophy patient organization in the SR financially supported also the training of three physiotherapists on correct sitting in the wheelchair – an issue which still marginalized and underestimated in Slovakia. These therapists are located in all regions (Bratislava – the west, Liptovský Mikuláš – centre, Košice – the east) and available as necessary to advise the members on the choice of a new wheelchair or to solve other disease related problems.

Thanks to another grant, the organization could equip its facilities with rehabilitation aids. Thus, the DystroCentrum project originated and it now provides free-of-charge consultation with physiotherapists not only to the members but also to other people with NMD (<http://www.omdvrs.sk/index.php/90-kat-aktuality/227-dystrocentrum.html>). It is necessary to promote this possibility and extend the target group so that the project can function on a long-term basis and benefit the community of NMD patients.

Over 12 years, the organization has become one of the subjects which organize public fundraisers and are perceived by the public as serious and well-established associations which help their members in a concrete way. Since the very beginning, public education on the disease and its consequences on people, their lives, and families has been a part of the campaign, which is symbolized by a blue butterfly. Although the fundraiser cannot be classified as a big one in terms of its extent and money collected, over the twelve years, the collected money has helped the members to buy appropriate and necessary compensation aids in 341 cases, totalling 23 881 912EUR. The members are not limited when applying for the help from the fundraiser – they can be helped several times, whenever there is a need to use an aid which compensates the consequences of disability.

**Recreational activities** under MDO in the SR have had many years' tradition and are popular with the members. They are sought after and they are all booked up shortly after the call for applications has been made.

The organization is particularly devoted to the camps for children with NMD, especially boys with DMD. It is an opportunity to target families with children suffering from this disease. The whole family, including the siblings, can come to the camp; therefore, the fee is only symbolic. MDO in the SR almost fully funds the camps and it obtains the funds from different sources. Currently, it is possible to financially contribute to the organization online through a donation portal (<http://www.ludialudom.sk/omd/>) by the end of March 2013. Every child in the camp has its own personal assistant provided by our organization. Assistants help the children during the whole camp programme. The programme is prepared by a special team of animators and it takes into account the health limitations of the children. A physiotherapist takes care of the children and teaches parents how to rehabilitate at home. A social counsellor and a psychologist are at parent's disposal to solve their problems. Parents have enough time to relax and build a self-help group; families keep in touch and build their relationships also after the end of the camp.

Activities organized every year also include integrative national meetings of members. A special programme is prepared for the participants, which is build on the latest information about the events within the organization, information from social and health

area (also in the form of counselling and seminars). The programme also includes cultural events, sport activities, space for members' experience sharing and self-help.

**The group of relief services** includes providing specialized social services by the Personal Assistance Agency founded under MDO in the SR. It provides wide-range service for 180 personal assistance users in Bratislava region; however, counselling covers the whole Slovakia. The clients appreciate and seek the support service of the agency; personal assistance enables them to live independently and significantly unburdens their families. The database of personal assistants helps the users of personal assistance to look for help with their day-to-day activities such as personal hygiene, help in the household, accompaniment, etc. The applicants are interviewed and the philosophy of the help and information on administration are discussed. The agency helps personal assistance users to arrange allowances and the related administration, communicate with assistants, and solve potential problems (<http://www.omdv.sr.sk/index.php/agentura-osobnej-asistencie/cinnost.html>).

All activities of the agency are funded from organization's sources. Neither the state nor cities are subsidizing these activities at the moment, in spite of the fact that the law on social services describes this service. However, the service is categorized as facultative.

For a long time, the organization has been making effort to change regulations in the legislation which concern several aspects essential for people with NMD.

Financing of the expenses for a protected workplace will be complicated after the law on employment services has been amended. The new law will introduce optionality for certain allowances; for some it will lower the support. This can lead to problems with sustainability of the expenses for protected workplaces and to reduction of the number of disabled employees. This will be reflected in a cut down on the extent of performed activities.

Income limit is a persistently unsolved problem of people who use personal assistance. The law on allowances for disability compensation specifies the limit of 4 multiple of the living wage. This causes decrease in the extent of help or potentially its withdrawal from personal assistance users. In a way, it forces disabled people to be passive recipients of state welfare. They are not motivated to be independent, because as soon as they reach certain income limit, all other expenses for assistance, which enables them to function independently, will have to be paid from their own income (in addition to standard expenses such as utilities, medication, food, etc.). However, the user, a disabled person, is not able to cover the real assistance costs from his salary.

Dystrophy patients have been criticizing the system of providing health aids from public health insurance and by means of commenting on legislation they have been trying to improve the accessibility of health aids. Unfortunately, despite the declared effort of the Ministry of Health of the Slovak Republic (hereafter MH SR) no practicable solution has been found so far. Dystrophy patients are dependent on several aids at once and the system has been consistently more or less ignoring this fact. The argument that dystrophy patients also need to use a mechanical wheelchair – some of them need a lightweight type – and have an electrical wheelchair, adjustable bed with anti-decubitus mattress at their disposal at the same time has not convinced MH SR representatives about the need to change the system.

It is also necessary to bring attention to the fact that the inherited and idiopathic neuropathy (G60), which belongs to rare NMD, is not included among diagnoses entitled to a spa treatment. The organization has been drawing attention of MH SR to this fact for several years, however, without any response. Yet, rehabilitation is often the only way how to slow down the progress of the disease. Another long neglected problem in the health care area is no reimbursement for an assistant during spa treatment. There are two aspects of this problem. Financial aspect – the health insurance company pays immobile patients' expenses for an assistant only symbolically; therefore, most dystrophy patients cannot afford spa treatment. The other aspect regards the accessibility – only a small number of spa facilities meet the requirements for barrier-free access, which in addition to entrance includes also barrier-free accommodation, lifts at spa procedures, adequate and trained assisting staff.

Slovakia is not doing enough to provide financial support for functioning of patient organizations – the subsidy system does not take them into account and grant support is non-existent. A tool for change could and should be the national plan for rare disease, which will not be really implemented without providing subsidies for patient organizations working in the area of rare diseases. In 2012, the organization became a member of EURORDIS and Alliance TREAT – NMD, which connects patients, doctors, scientists and pharmaceutical companies in the field of NMD research.

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## **PRAKTICKÉ SKÚSENOSTI V POSKYTOVANÍ ŠPECIÁLNYCH SOCIÁLNYCH SLUŽIEB PRE PACIENTOV SO ZRIEDKAVÝMI CHOROBAMI**

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Organizácia muskulárnych dystrofií v SR, Slovensko

Organizácia muskulárnych dystrofií v SR (OMD v SR) je jediná špecifická organizácia na Slovensku, ktorá združuje deti a dospelých so svalovou dystrofiou a iným druhom nervovosvalového ochorenia (ďalej NSO) a ich rodiny. Členom s týmito zriedkavými chorobami poskytuje nielen ľudské zázemie, ale aj odbornú pomoc.

Organizácia vznikla ako samostatné občianske združenie v roku 1993. Kontinuálne poskytuje sociálne poradenstvo a ďalšie špecializované sociálne služby pre členov a klientov. V počiatočných rokoch vykonávala svoju verejnoprospešnú, neziskovú činnosť na dobrovoľníckom základe, dnes sa na jej aktivitách podieľa deväťčlenný profesionálny tím – siedmi zamestnanci s ťažkým zdravotným postihnutím (ďalej ŤZP) a dvaja pracovní asistenti. K tejto zmene mohlo prísť predovšetkým vďaka získaniu štatútu chráneného pracoviska v roku 2009. V súlade s odporúčaniami EURORDIS zastrešuje Organizácia muskulárnych dystrofií v SR aktivity v 3 hlavných oblastiach: informačný servis (*information services and help line*), rekondičné aktivity (*therapeutic recreational programmes*) a odľahčovacie služby (*respite care services*). V rámci informačného servisu poskytuje v pracovné dni špecializované sociálne poradenstvo pre ľudí s ŤZP telefonicky, e-mailom, osobne alebo terénnou formou v domácnosti klienta.

Organizácia tiež vydáva vlastný, kvalitný časopis Ozvena, ktorý štyrikrát do roka bezplatne distribuuje každému členovi až domov. Na jeho tvorbe sa podieľajú samotné osoby s NSO z pozície odborníkov aj pacientov. Pre mnohých členov je to stále jediný zdroj informácií o ochorení a s tým súvisiacich problémoch. Organizácia sa špeciálne venuje detským táborom pre deti s NSO, hlavne chlapcom s DMD. Každoročne organizované aktivity zahŕňajú tiež interaktívne národné stretnutia členov.

Skupina odľahčovacích služieb zahŕňa poskytovanie špecializovaných sociálnych služieb v Agentúre osobnej asistencie, ktorá bola založená pod hlavičkou OMD v SR. Zabezpečuje široký servis pre 180 užívateľov osobnej asistencie v Bratislavskom kraji, poradenstvom však pokrýva celé Slovensko.

Organizácia sa dlhodobo snaží o zmenu nastavení resp. legislatívy v niekoľkých pre ľudí s NSO zásadných otázkach.

Veľké rezervy má Slovensko v poskytovaní finančnej podpory na činnosť patientskych organizácií zo strany MZ SR, nakoľko dotačný systém pre ne nie je nastavený vôbec a grantová podpora je nulová. Nástrojom na zmenu by mohol a mal byť práve národný plán starostlivosti o zriedkavé choroby, ktorého skutočná implementácia nenastane bez spustenia poskytovania dotácií pre patientske organizácie pôsobiace v oblasti vzácnych ochorení. V roku 2012 bola organizácia prijatá za člena EURORDIS a Aliancie TREAT – NMD, ktorá spája pacientov, lekárov, vedcov a farmaceutické firmy na poli výskumu NSO.

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