

WITH FUTURE IN MIND

Neuromuscular disorders (NMD) are disease of motor unit with muscular weakness as predominant and unifying clinical sign. They can affect people of all age groups. In NMD we traditionally count hereditary disorders of muscle like muscular dystrophies and congenital myopathies, disorders of neuromuscular junction like myasthenia gravis, disorders of nerves like hereditary motor and sensory neuropathies, and disorders of neurones like spinal muscular atrophies. NMD are usually chronic progressive disorders that can often lead to severe physical impairment.

NMD are relatively rare. Total number of patients with NMD in Slovenia (2 000 000 inhabitants) in national register (Centre for neuromuscular disorders, Neurological clinic, Clinical centre Ljubljana) does not exceed 1200.

Yet, NMD are important group of diseases. In a way they are a prototype of genetic disorders. They are also scientific and clinical challenge. A patient with NMD often needs well organised and fast responding health service. From general practitioner or neurologist she or he expects to recognise the disease, from geneticists the proper counselling, from orthopaedic surgeon to treat scoliosis, from internal medicine specialist to recognise and treat myocardiopathy or ventilation problems and from rehabilitation team to help to provide the technical aids and address the handicap. Patients with NMD with their needs and medical problems are continuously testing expertise and organisation of every health system and also moral values of whole and each society all over the globe.

Therefore, it is no surprise that associations of patients have been formed early by the patients with NMD to influence the social milieu especially social, educational and health systems. Društvo distrofikov Slovenije (Slovene Muscular Dystrophy Association) has been founded in 1970. Since then it has been an active promoter of awareness and fight for the rights of people with disabilities. As a part of the Yugoslav MDA it has also been an active co-founder of EAMDA (European Association of Muscular Dystrophy Associations).

Slovene Muscular Dystrophy Association has welcomed the 32nd Annual General Meeting of EAMDA from 26th to 29th September 2002 in Ljubljana, Slovenia. Within the meeting the two day scientific *Symposium on Biological and Social Resources for the Advancement of the Quality of Life of People with Neuromuscular Disorders* has been held.

Slovenia has a rich scientific and clinical tradition in the field of NMD. The knowledge and experience have been gained through the co-operation between different disciplines and institutions being medical (Clinical Centre in Ljubljana, Rehabilitation Institute, Medical Faculty in Ljubljana) or technical (Jožef Stefan Institute, University in Ljubljana). Co-operation with technical expertise promises excellent opportunities for progress in the field of rehabilitation medicine also for people with NMD. Part of these fast expanding and attractive possibilities was presented at the meeting.

Slovene model of management of people with NMD is, as we believe, modern and could stand the test of comparison with the best practices in the world. Slovenia is a pioneer in the rehabilitation (restorative neurology) of patients with NMD. We can be proud of achieved but there is much to be done.

The path to the cure is long and equipped with many curves and obstacles but every inch along that way provides new important discoveries and adds new knowledge. While taking that route we must not forget the patients with NMD who need our help here and now. We asked ourselves what has been found new in science and clinical practise that could be implemented in the better care for people with NMD.

One may find most of the contributions to the *Symposium on Biological and Social Resources for the Advancement of the Quality of Life of People with Neuromuscular Disorders* in this supplement of Zdravniški vestnik. We thank all who have made it possible.

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Z MISLIMI NA PRIHODNOST

Živčnomišične bolezni (ŽMB) so bolezni motorične enote. Njihov prevladujoči in skupni klinični znak je mišična šibkost. ŽMB prizadenejo vse starostne skupine. V to skupino tradicionalno prištevamo večinoma dedne bolezni mišic, kot so npr. mišične distrofije in kongenitalne miopatije, bolezni živčnomišičnega stika, kot je npr. miastenija gravis, bolezni živcev, kot so npr. dedne motorične in senzorične polinevropatije, in nevronopatije, kot je npr. spinalna mišična atrofija. Praviloma gre za kronične napredujoče bolezni, ki pogosto vodijo do hude gibalne prizadetosti.

Ljudi s posamezno obliko živčnomišične bolezni je sorazmerno malo. Skupno število zabeleženih pacientov v sicer neobveznem registru Centra za živčnomišične bolezni, SPS Nevrološka klinika Kliničnega centra v Ljubljani, v Sloveniji ne presegata številke 1200.

Pa vendar gre za pomembno skupino bolezni. ŽMB predstavlja prototip genetskih bolezni. So znanstven kot klinični izziv. Pacient z ŽMB pogosto potrebuje organizirano oskrbo več služb. Tako npr. od splošnega zdravnika in nevrologa pričakuje, da bosta prepoznala bolezen, od genetika svetovanje, od ortopeda, da bo skušal popraviti skoliozo, od internista, da bo zdravil miokardiopatijo in restriktivne motnje dihanja, ter od fiziatra, da mu bo pomagal z ustreznimi tehničnimi pripomočki in rehabilitacijskimi metodami zmanjšal oviranost. Potrebe pacientov z ŽMB zato neprestano preverjajo zdravstveni sistem in celotno družbo po vsem svetu (spomnimo se javne polemike o ALS pri nas). Po drugi strani vse bolj razumemo patološke procese, ki privedejo do izražanja ŽMB, kot predčasno staranje struktur motorične enote (glej prispevek Vrbove in Hausmanow v nadaljevanju). Zato upamo, da bodo znanje in postopki v procesu zdravljenja ŽMB pomembno vplivali tudi na razumevanje staranja in na postopke zdravljenja tegob ljudi v takojmenovani starosti. Problemi ŽMB so torej pereči raziskovalni problemi sodobne razvojne in molekularne biologije.

Tako ni čudno, da so se ljudje z ŽMB med prvimi povezali v društva, da bi vplivali na družbeno okolje, zlasti na socialne, izobraževalne in zdravstvene sisteme. Društvo distrofikov Slovenije je bilo ustanovljeno 1971. Vse od takrat je dejavni nosilec ozaveščanja in borbe za pravice ljudi s posebnimi po-

trebami. Bilo je, takrat še najživahnejše društvo v SFRJ, tudi pobudnik ustanovitve Evropske zveze društev ljudi z ŽMB (EAMDA).

Šestindvajsetega do devetindvajsetega septembra 2002 je Društvo distrofikov Slovenije gostilo Dvaintrideseto letno zasedanje EAMDA-e (32nd Annual General Meeting of EAMDA) v Ljubljani. V okviru srečanja smo na pobudo Društva distrofikov Slovenije pripravili dvodnevno strokovno srečanje z naslovom *Simpozij o bioloških in socialnih virih za izboljšanje kakovosti življenja ljudi z živčnomišičnimi boleznimi (Symposium on Biological and Social Resources for the Advancement of the Quality of Life of People with Neuromuscular Disorders)*.

Slovenija ima bogato tradicijo kliničnega in znanstvenega dela na področju ŽMB. Znanje in izkušnje smo pridobili tudi v sodelovanju med različnimi strokami in ustanovami tako medicinskimi (Klinični center, Inštitut Republike Slovenije za rehabilitacijo, Medicinska fakulteta) kot tehničnimi (Fakulteta za elektrotehniko, Inštitut Jožef Stefan). Sodelovanje s tehničnimi strokami kaže izredne možnosti napredka. Del teh možnosti so sodelavci predstavili na srečanju.

Mirno lahko zatrdimo, da je slovenska zasnova obravnave ŽMB moderna in v vseh pogledih sledi najboljšim modelom v svetu. Zlasti na področju rehabilitacijske medicine, ki jo temeljimo na načelih restorativne nevrologije, je Slovenija med pionirji. Zagotovo pa si želimo boljše organizacijske podpore.

Vsi, zlasti seveda oboleli z ŽMB, željno pričakujemo zdravlilo. Do njega nas vodi zavita pot polna novih odkritij in spoznanj. V pričakovanju možnosti zdravljenja dednih bolezni pa ne smemo pozabiti na ljudi z ŽMB, ki potrebujejo pomoč tu in zdaj. Med pripravami na posvet smo se vprašali, kaj je prinesla znanost ali klinična praksa novega, kar bi lahko utemeljeno uporabili na tem področju in tako pomagali ljudem z ŽMB.

Večino prispevkov srečanja *Simpozij o bioloških in socialnih virih za izboljšanje kakovosti življenja ljudi z živčnomišičnimi boleznimi* boste našli v pričujoči posebni številki Zdravniškega vestnika.

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