

Resolution of International Conference
“Recent standards in diagnosis, treatment and medical support for some rare neuromuscular diseases”

An international research and practical Conference entitled “Recent standards in diagnosis, treatment and medical support for some rare neuromuscular diseases”, organized by “Institute of Neurology, Psychiatry and Narcology AMS of Ukraine” with participation of Kharkiv Foundation «Children with spinal muscular atrophy» was carried out in Kharkiv on 21-23 May 2009.

Leading researchers of Ukraine, Great Britain, Sweden, and France, specialists in neurology, psychiatry, orthopedics, rehabilitation, pulmonology, as well as parents and relatives of patients participated in the Conference.

Researchers and general practitioners, members of parents’ organization Kharkiv Foundation «Children with spinal muscular atrophy» realize that the nation’s health in general and the life of patients with rare genetic diseases, including spinal muscular atrophy (SMA), Duchenne muscular dystrophy (DMD) and other rare neuromuscular diseases (rNMDs) depends on scientific achievements and joint efforts of both state and parents. The goal of the participants is consolidation of creative potential for knowledge deepening, experience exchange, elaboration of ways for implementation the most up-to-date treatment, diagnostics, and prophylaxis of SMA, DMD and other rare neuromuscular diseases.

Contemporary aspects of diagnostics, treatment, prophylaxis and rehabilitation of patients, patient care, registration of patients, ethical aspects of interrelations between physicians and patients, directions and perspectives of international cooperation were discussed during the Conference.

It has been noted that neuromuscular diseases are a group of inherited degenerative diseases with progressive course of pathological process. General list of rNMDs includes up to 300 different conditions, determined as rare, with prevalence less than 5:10’000 of population, or 1:2’000 , 1:6’000 etc. according to other authors. Most of these disorders are of chronic course, present the reason for severe disability of patients, and become significant burden for the society.

The problem of aiding rNMDs patients is complicated by the fact that this disease is diagnosed in both newborns and adults.

Primary diagnosis of the disease is rather complicated due to the disease manifestation through the syndromes characteristic of other known diseases – arrested physical and psychic development, epileptic, myopathic and other symptoms, as well as prevalence of genetic heterogeneity over specific clinical symptoms. Verification of most rNMDs requires special laboratory equipment, reagents, specific DNA diagnostic methods, complicated chromatographic methods, as well as specialists experienced in rNMDs.

Treatment of rNMDs patients presents another challenge. Different methods of medicamental, orthopedic, physiotherapeutic and psychotherapeutic treatment of such patients exist. Nevertheless, all of them are symptomatic.

Etiopathogenetic therapy of this group of patients is not available for the moment, and the main goal of medical surveillance is minimization of effect of such complications as deformity of spine and joints, maintenance of cardiac and respiratory function.

Legislative base and state statistical registration of rNMDs is deficient, no systematic approach to treatment and medicamental provision of such patients is implemented.

Participants of the Conference have attracted attention of the society, health care professionals and parents to the fact that the conditions for effective realization of actual aid either within Ukraine or in the frame of international cooperation are impossible to create without high quality diagnostics, patient registration system, and participation of the state in solution of existing problems and adequate attention to the patients with SMA, DMD, rNMDs.

“Institute of Neurology, Psychiatry and Narcology AMS of Ukraine”, Ukrainian medical centre of rehabilitation children with organic defeat of CNS, Donetsk pediatric regional centre of neuro-rehabilitation, Kharkiv Foundation «Children with spinal muscular atrophy» have accumulated the experience in solution of such problems.

Joint efforts of state authorities, scientists, general practitioners and parents are the only source of social effect in the sector of aiding patients with SMA, DMD, rNMDs.

Participants of the Conference, having discussed the reports presented and determined the most urgent problems, have arrived to the following conclusions:

1. Progressive neuromuscular diseases should be construed an important medico-social problem.
2. State medical statistical registration of SMA, DMD and other rNMDs patients requires improvement.
3. Research teams together with healthcare professionals should elaborate the standards of diagnostics, treatment, rehabilitation, prophylaxis, and aiding rNMDs patients.
4. Coordination program should be elaborated, and the scope of healthcare professionals' activity, i.e. of pulmonologists, orthopedists, prosthetics specialists, and neurologists in aiding SMA, DMD, rNMDs patients should be determined. These activities are expected to promote both prolongation of life span of the patients suffering from the mentioned disorders and improvement of their quality of life.
5. Conditions for improvement of rNMDs diagnostics and diagnosis verification both within Ukraine and by consulting European diagnostic centers should be created.
6. Organization and establishment of an interdepartmental committee for coordination of activities according to SMA program for effective and operative interaction between state authorities, researchers, parents' organizations and international community should be promoted.
7. Training programs of medical universities and academies of postgraduate education should include the questions of diagnostics and early recognition of rNMDs patients of different age.
8. Legislative base of aiding rNMDs patients and their medicamental provision should be elaborated.
9. The problem of organization of a specialized rare neuromuscular disease center as a leading institution coordinating the activity of all the institutions and organizations concerned should be discussed.
10. rNMDs questions should be included to the plans of research activities.
11. Research and practical Conferences and working meetings devoted to the problems of SMA, DMD and other rNMDs should be carried out regularly.

ORGANIZING COMMITTEE OF THE CONFERENCE