NMD SerbNet

SERBIAN NEUROMUSCULAR DISEASE NETWORK

Improving diagnostics, enhancing research and advancing therapy of neuromuscular diseases in *Serbia* through well planned long-term improvement and introduction of innovative practices in all aspects of neuromuscular diseases

GOALS OF THE NETWORK

Systematisation of clinical data, molecular genetic analyses data, information on availability of biological samples, as well as data collected from scientific research activity related to Serbian patients suffering from neuromuscular diseases

Increasing the number of neuromuscular diseases whose complete diagnosis can be performed in Serbia through adoption of standard procedures and state-of-the art diagnostic protocols for neuromuscular diseases

Completion of national patient registries and their inclusion in global registries

Increasing the number of biological samples stored in the national biobank and joining the European network of biobanks – The EuroBioBank

Recruitment of clinicians and researchers from Serbia involved in neuromuscular diseases

Strengthening the interaction with patient organisations

Strengthening existing and establishing novel collaborations with regional and international centres for neuromuscular diseases

Active participation in international projects concerning neuromuscular diseases

NMD SerbNet

Clinic for Neurology and Psychiatry for Children and Youth,
Faculty of Medicine, University of Belgrade
https://sites.google.com/site/decijaneuropsihijatrija/Home

prof. dr Vedrana Milić Rašić, executive board member
dr Ana Kosać
dr Jelena Nikodinović Glumac
dr Jelena Mladenović

Neurology Clinic, Clinical Centre of Serbia,
Faculty of Medicine, University of Belgrade
http://neurologija.bg.ac.rs

prof. dr Vidosava Rakočević Stojanović, executive board member
dr Stojan Perić

Clinical Hospital Centre Zemun
www.kbczemun.rs

prof. dr Sanja Milenković, executive board member

Centre for Human Molecular Genetics,
Faculty of Biology, University of Belgrade
www.dnkanaliza.rs

prof. dr Dušanka Savić Pavićević, executive board member
Miloš Brkušanin
Jovan Pešović

Institute for Human Genetics,
Faculty of Medicine, University of Belgrade
http://www.mfub.bg.ac.rs

prof. dr Ivana Novaković, executive board member

Institute for Molecular Genetics and Genetic Engineering,
University of Belgrade
www.imgge.bg.ac.rs

dr Snežana Kojić, network coordinator
dr Dragica Radojković, executive board member
dr Aleksandra Nestorović
Jovana Jasnić Savović

Supported by:

NMD SerbNet

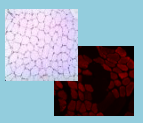
Accurate diagnosis of neuromuscular diseases can be extremely complex and challenging given the fact that many of these diseases have overlapping symptoms. Following a comprehensive clinical examination, genetic testing and/or a muscle/nerve biopsy might be requested.

Clinical examination consists of a thorough neurological examination, laboratory tests (biochemical, immunological, etc.), neurophysiological tests (electromyoneurography, EMNG), ultrasound scan and radiology tests (RTG, CT, MR). These tests can be carried out at *Clinic for Neurology and Psychiatry for Children and Youth, Faculty of Medicine, University of Belgrade* and *Neurology Clinic, Clinical Centre of Serbia.*

Genetic testing allows the unequivocal diagnosis of certain neuromuscular diseases, as well as prenatal testing of individuals with family history of neuromuscular diseases. Application of modern techniques for the detection of different types of DNA mutations enabled diagnosis of around twenty inherited neuromuscular diseases in Serbia, among which are the most common forms of muscular dystrophies, motor neuron diseases, hereditary neuropathies and mitochondrial diseases (Table). Genetic testing is offered by two institutions: *Centre for Human Molecular Genetics, Faculty of Biology* and *Neurology Clinic, Clinical Centre of Serbia.*

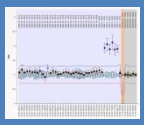
Biopsies in adult patients are taken at *Clinical Hospital Centre Zemun*, while *University Children's Hospital* performs biopsies in children. All biopsies are scheduled in advance (contact person is prof. Sanja Milenković, tel. +381648543071). Tissue sample is analysed in the histopathology laboratory using standard histological and enzyme histochemical techniques. A fraction of the tissue sample is stored in the Serbian biobank for neuromuscular diseases located at *Institute of Molecular Genetics and Genetic Engineering.*

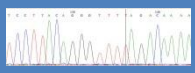
CONTACT
dr Snežana Kojić, network coordinator
snezanakojic@imgge.bg.ac.rs

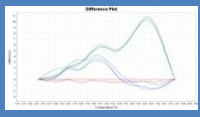


NMD SerbNet

Disease	Gene/Type of analysis	Laboratory
Duchenne and Becker Muscular Dystrophy (DMD/BMD)	DMD duplications and deletions	CHMG/NC
	DMD haplotype analysis	CHMG
Myotonic dystrophy (DM1)	DMPK	CHMG, NC
Myotonic dystrophy (DM2)	CNBP (ZNF9)	CHMG
Limb girdle muscular dystrophy 2A (LGMD2A)	CAPN3	NC
Hereditary Motor and Sensory Neuropathies (Charcot-Marie-Tooth, CMT)	PMP22 duplication	CHMG, NC
	PMP22 sequencing	CHMG
	MPZ	
	GJB1	
	LITAF (SIMPLE)	
	EGR2	
	NDRG1	
Neuromyotonia and Axonal Neuropathy (NMAN)	HINT1	CHMG
Congenital Cataracts, Facial Dysmorphism and Neuropathy (CCFDN)	CTDP1	CHMG
Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)	PMP22 deletion	CHMG, NC
Congenital Myasthenic Syndrome (CMS)	CHRNE	CHMG
Spinal Muscular Atrophy (SMA)	SMN1	CHMG
Amyotrophic Lateral Sclerosis 1 (ALS1)	SOD1	CHMG
	TARDBP	
	ANG	
Frontotemporal Dementia and/or Amyotrophic Lateral Sclerosis (FTDALS1)	C9orf72	CHMG, NC
Kennedy's disease (SBMA)	AR	CHMG, NC
Mitochondrial disorders	mtDNA sequencing	NC
Myoclonic Epilepsy with Ragged-red Fibers (MERRF)	MT-TK	CHMG







CHMG

Centre for Human Molecular Genetics,
Faculty of Biology, University of Belgrade
contact person: prof. dr Dušanka Savić Pavićević
duska@bio.bg.ac.rs

NC

Neurology Clinic, Clinical Centre of Serbia,
Faculty of Medicine, University of Belgrade
Contact person: prof. dr Ivana Novaković
tetaana61@yahoo.com