

Academic Days of the National Clinical Center for Children's Neuropsychomotor
Recovery "Dr. Nicolae Robănescu", Bucharest, 14-15 September 2023
- Congress Abstracts

Conference Abstract – L03

The Importance Of A Fast And Accurate Diagnosis In Lysosomal Diseases

Elena-Silvia Shelby¹, Andra Pintilie¹, Liliana Padure^{1,2}, Mihaela Axente^{1,3}, Elena Neagu¹, Anca Irina Grigoriu¹, Nicoleta Chiriloi¹, Corina Sporea^{1,2}, Andrada Mirea^{1,2},

1. „Dr. Nicolae Robănescu” National Clinical Center for Children’s Neurorehabilitation, Bucharest
2. Chair of Balneofiziokinetotherapy and Rehabilitation, Faculty of Midwifery and Nursing, „Carol Davila” University of Medicine and Pharmacy, Bucharest
3. Chair of Pathophysiology II, Faculty of General Medicine, „Carol Davila” University of Medicine and Pharmacy, Bucharest

Lysosomal diseases are storage disorders caused by cellular dysfunction due to the accumulation of the substrate in the context of lysosomal malfunction. We present two cases of lysosomal diseases from Dr Nicolae Robănescu National Clinical Center for Children’s Neurorehabilitation – a 17-year old boy diagnosed with mucopolysaccharidosis type A and an 8-month old boy diagnosed with Krabbe disease. A fast and accurate diagnosis of lysosomal diseases is mandatory, as currently there is treatment for some of these diseases, such as Gaucher, Fabry, Pompe, various types of mucopolysaccharidoses, and others.

Conference Abstract – L04

Multifactorial Motion Analysis In The Diagnostic And Therapeutic Approach Of The Gait Disorders In Children With Spastic Hemiparesis

Raluca Petcu¹, Anca Grigoriu²,

1. Municipal Hospital of Turnu Magurele
 2. National Clinical Center of Neurological Rehabilitation for Children “Dr. Nicolae Robănescu”, Bucharest
- Corresponding author: Irina Raluca Petcu , E-mail: varsa_raluca@yahoo.com

Abstract

Introduction: Spastic hemiparesis represents 30% of all forms of cerebral palsy (CP). Of these, 96% acquire the walking ability, 90% of which independently. The individualization of the therapy, surgery and/or rehabilitation, for the children with spastic hemiparesis is a key element in the therapeutic approach.

The Multifactorial Gait Analysis (MGA) represents an instrumental (objective) method of the gait pattern assessment using advanced integrated technologies (GAITLAB). It allows the identification of the kinematic patterns in spastic hemiparesis (congenital or acquired) and their classification (1).

Objective: Through this work, we aimed to highlight the way of individualization and adaptation of the therapy depending on the gait pattern and the intrinsic changes following the MGA in the children with spastic hemiparesis.

Material and methods: Examples were taken from the casuistry of the GaitLab of the National Clinical Center of Neurological Rehabilitation for Children „Dr. Nicolae Robănescu”, regarding patients with congenital or

acquired spastic hemiparesis and the specific therapeutic characteristics (rehabilitation, botulinum toxic injections, orthotics, surgery) for each gait pattern identified.

Discussions and conclusion: The MGA is an important, precise and useful method for evaluating gait disorders, which facilitates and improves the therapeutic approach in children with congenital or acquired spastic hemiparesis (2).

Bibliography:

1. J Rodda, H K Graham. Classification of gait patterns in spastic hemiplegia and spastic diplegia: a basis for a management algorithm. Eur J Neurol dec. 2001.
2. J. Gage, M. Schwartz, S. Koop & T. Novacheck (Eds.), The identification and treatment of gait problems in cerebral palsy. London: Mac Keith Press. 2009

Conference Abstract – L08

Approaches And Achievements Of The Romanian Society Of Physical Medicine, Rehabilitation And Balneoclimatology (RSPMRB) And Romanian Perspectives In This Field, Including Aiming At The Amplification Of International Collaborations

Prof. Gelu ONOSE, MD, PhD, MSc; Assist. Prof. Cristina POPESCU, MD, PhD; Assoc. Prof Dr. Biol. Constantin MUNTEANU; Assoc. Prof. Dan BLENDEA, MD, PhD.

Introduction

The Romanian Society of Physical Medicine, Rehabilitation and Balneoclimatology (RSPMRB) represents a professional-scientific entity whose overall goal is to improve the quality of health services in the field of Physical Medicine, Rehabilitation and Balneology (PMRB) including through a sustained academic activity. Our presentation contains highlights of the main efforts and achievements of the RSPMRB – established in 2015 and affiliated to the International Society of Physical and Rehabilitation Medicine (ISPRM) in 2020 – realized through a very consistent but also valuable contribution to the development of this field.

Material and method

The presentation will chronologically highlight the most important milestones in the activity of RSPMRB, the organization of numerous scientific events at national and international level - starting with the 41st World Congress of the International Society of Medical Hydrology and Climatology and, respectively, of Physical Medicine, of Rehabilitation and Balneology, held in Bucharest in 2016 - as well as an appreciable number of important works, editorial appearances, at national and international level, as well as a constant promotion and facilitation of professional dialogue.

Result

The SRMFRB constituted and still constitutes a space for dialogue and constructive collaboration between specialist doctors - and from specialties closely related to the field of MFRB - physio-/kineto-therapists but also the middle staff working in this profile and not least with the representatives of the National House of Social Health Insurance, achieving significant results in improving the funding level of this field.

Discussions

SRMFRB's activity has also known: constancy and continuous progress in performance and efficiency, including managing to maintain high standards of performance and concrete achievements - in the sense of the above - including overcoming the difficulties imposed by the COVID-19 pandemic.

Conclusion

The consistency of multi-level achievements of SRMFRB in the almost 8 years of its existence also represents a particularly solid basis of trust for continuity and development in performance and professional-academic effectiveness.

Conference Abstract – L11

The Influence Of Demographic Variables On Quality Of Life: A Comparison Of Families With Children Who Have Acquired Versus Congenital Conditions.

M. V. Morcov^{1,2}, L. Padure^{1,2}, C. G. Morcov¹, G. Onose^{2,3}

Affiliations:

1. „Dr. N. Robanescu” National Clinical Center of Neurorehabilitation for Children, Bucharest, Romania
2. Faculty of Medicine „Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania
3. The Neuromuscular Rehabilitation Clinic Division „Bagdasar-Arseni” Clinical Emergency Hospital, Bucharest, Romania

The review of the literature shows that there are many studies that analyzed socio-demographic parameters: education, marital status, place of residence and income and reported to QOL in families with children with disabilities, but no comparative statistical analyses, carried out between families with congenital vs. acquired neurodisabilities [1,2].

The study, approved by the Ethics Commission (No. 8739/ 28.10.2019), was conducted in the NTCNRC “Dr. N. Robanescu” and included 152 mothers of children and adolescents (87 mothers of patients with congenital neurodisabilities – Group 1 and 65 mothers of patients with acquired neurodisabilities – Group 2) who were questioned using the PedsQL- Family Impact Module (PedsQL-FIM) [3] and the World Health Organization Quality of Life Instrument, Short Form (WHOQL-BRIEF) [4], Romanian versions.

According to our data, there were no correlation between socio-demographic parameters and used questionnaires for Group 1 and a weak, negative correlation between physical health and marital status (WHOQL-BRIEF questionnaire) for Group 2.

Further research is needed with an increased number of participants and, consequently, higher statistical power for a deeper analysis.

References

1. Vilaseca RM, Rivero M, Bersabé RM, Cantero MJ, Navarro-Pardo E, Valls-Vidal C, et al. Demographic and parental factors associated with developmental outcomes in children with intellectual disabilities. *Front Psychol* 2019;10. <https://doi.org/10.3389/fpsyg.2019.00872>.
2. Roddy Á. Income and conversion handicaps: estimating the impact of child chronic illness/disability on family income and the extra cost of child chronic illness/child disability in Ireland using a standard of living approach 2022;23:467–83. <https://doi.org/10.1007/s10198-021-01371-4>.
3. Varni, J.W.; Sherman, S.A.; Burwinkle, T.M.; Dickinson, P.E.; Dixon, P. The PedsQLTM Family Impact Module: Preliminary reliability and validity. *Health Qual. Life Outcomes* 2004, 2, 55.
4. Garnefski, N.; Kraaij, V. The cognitive emotion regulation questionnaire: Psychometric features and prospective relationships with depression and anxiety in adults. *Eur. J. Psychol. Assess.* 2007, 23, 141–149.

Conference Abstract – L12

The Increased But Unmet Needs Of Neurofibromatosis Type 1

Autori: Nicoleta Chiriloi¹, Leanca Madalina^{1,2}

¹ National University Hospital for Children Neurorehabilitation "Dr. Nicolae Robănescu" (NUHCN)

² University of Medicine and Pharmacy "Carol Davila"

Introduction. Neurofibromatosis type 1 (NF1) is a rare condition with autosomal dominant transmission diagnosed based on clinical diagnostic criteria. Due to the variety of expression, the systemic manifestations are diverse, and some signs are present from birth, while others appear over time. Plexiform neurofibromas (PN) occur in ~30%–50% of patients with NF1 and are peripheral nerve tumors arising from multiple nerve fascicles. These PNs can lead to facial deformities, motor disorders, pain, respiratory disorders, visual disturbances, and bladder/bowel disorders. PNs are inoperable when they cannot be completely removed without risk of significant injury due to location around or in proximity to vital structures, invasiveness, or increased vascularity of NPs in NF1. The first drug approved for the treatment of inoperable symptomatic PN from NF1 for children and adolescents, aged 3 years and older, is Selumetinib available in our country from 2021.

Objectives. Clinical examinations and periodic multidisciplinary monitoring help to identify early deficiencies that can be corrected leading to the reduction of severe complications of the disease, and the administration of Selumetinib, a selective inhibitor of MEK 1/2 that blocks the proliferation and survival of tumor cells in which the RAF pathway is activated - MEK-ERK allows tumor reduction of inoperable PNs with the improvement of children's quality of life.

Material and methods. The paper aims to present the results of treatment with Selumetinib in the case of a patient treated between 2021-2023 within the Compassionate Use Program in the National University Hospital for Children Neurorehabilitation "Dr Nicolae Robanescu" (NUHCN). The initiation of the treatment and subsequent clinical, biological and imaging monitoring was carried out within the multidisciplinary team within the NUHCN represented by a neurologist, pediatrician, orthopedist, psychiatrist, recovery and physical medicine physician, radiologist, cardiologist, anesthesiologist and intensive care physician, endocrinologist, geneticist.

Results. We observed the reduction in size of the PN volumes, as well as the modification of the structure and consistency of the PN with the help of 3 Tesla nuclear magnetic resonance imaging performed in multiple regions (cerebral, medullary). Also, the patient's quality of life through the volumetric reduction of PN was improved by the significant reduction of pain, motor disorders, and facial deformities. Also, the adverse reactions were monitored by the multidisciplinary team.

Conclusions. Selumetinib is the first drug approved for inoperable, symptomatic NF 1 PN that is well tolerated by children with positive effects by reducing the morbidity determined by the tumor volume. In the future, in-depth research is needed for the genotype-phenotype interrelationship regarding the response to the selective MEK 1/2 inhibitor treatment.

Keywords: neurofibromatosis, plexiform neurofibroma, selumetinib

Conference Abstract – L13

„Dr. N. Robanescu” National Clinical Center of Neurorehabilitation for Children – an Excellence Center

Liliana Padure^{1,2}, Corina Sporea^{1,2}, Maria Veronica Morcov^{1,2}, Cristian Gabriel Morcov¹

Affiliations:

1. „Dr. N. Robanescu” National Clinical Center of Neurorehabilitation for Children (CNCRNC), Bucharest, Romania
2. Faculty of Medicine, „Carol Davila” University of Medicine and Pharmacy (UMFCD), Bucharest, Romania

In 1968, Dr. Robanescu developed Romania's first functional rehabilitation unit for kids with physical and neurological disabilities.

The CNCRNC gains legal personality in 2007 and becomes integrated into the Ministry of Health of Romania. Our Center, which is affiliated with UMFCD, is now the only university clinic for pediatric rehabilitation in Romania, functioning as a training ground for all pediatric rehabilitation experts.

We provide training programs for doctors through intense courses in botulinum toxin injection, and we offer practical internships for UMFCD students. Conferences, Symposia, Workshops, Courses, etc. are all organized by CNCRNC, a provider of Continuing Medical Education.

The CNCRNC introduced the Nusinersen treatment for Spinal Muscular Atrophy for the first time in Romania in 2018, also being the provider of the new therapies for other pathologies.

We have modern robotic technology, which completes the traditional therapy approach to patients.

The National Health Quality Management Authority classified the Center in 2023 as a "Accredited Category II".

Conference Abstract – L14

Neurological Complications Secondary To Drug Consumption; Clinical And Pathophysiological Aspects

Assoc.Prof. Dr. Anghelescu Aurelian^{1,2}

1. Universitatea de Medicină și Farmacie "Carol Davila"
2. Clinica de Neurorecuperare, Spitalul Clinic de Urgenta "Bagdasar-Arseni", Bucuresti

Like many other countries, drug experimentation and consumption among young people and adolescents in Romania represent socio-medical and medico-legal challenges.

The paper is part of ongoing efforts to educate young people about the potential risks of various illegal drugs. Pathophysiological mechanisms of dependence and addiction are presented. Modern imaging methods (MRI, tractography, PET, SPECT) reflect the metabolic and structural changes induced in the brain by drug use.

Cannabis is among the most frequently consumed illicit drugs among teenagers in Romania. It is often (erroneously) seen as relatively affordable and less harmful by some individuals.

In addition to the "classic" illegal drugs (cocaine, heroin, meth), the emergence of synthetic drugs and new psychoactive substances (NPS) has been a significant challenge. These substances often mimic the effects of traditional drugs but can be more dangerous due to their unpredictable composition and potential health risks. The paper highlights the tremendous influence of the internet and social media in shaping perceptions and behaviors, including drug use. Online platforms can provide useful medical information and education, but they can also facilitate illegal drug transactions, making it essential for authorities to monitor and address this issue.

The paper also addresses legislation and law enforcement elements to combat drug trafficking and distribution. However, the effectiveness of these efforts can vary, and a balanced approach is needed to address supply and demand factors.

A key aspect is education, awareness, and prevention of illegal drug use and abuse: medical academic institutions, various organizations, and government agencies must work to raise awareness of the dangers of

drug use among young people. School programs, community awareness campaigns, and media campaigns can be used to inform adolescents about the potential consequences of drug use.

Conference Abstract – L15

Integrating The One Health Approach In Zoonotic Diseases Monitoring And Prevention. Lessons From The COVID-19 Pandemic

Authors: Dr. Văcaru Gabriel - Cristian^{1,2}, Dr. Oana Codruța Băcean Miloicov^{3,4}, Prof. Dr. Emanoil Ceaușu^{1,2,5}

1. Victor Babeș Clinical Hospital for Infectious and Tropical Diseases, Bucharest, Romania
2. Carol Davila University of Medicine and Pharmacy, Bucharest, Romania
3. "Victor Babeș" University of Medicine and Pharmacy, Department of Microbiology, Discipline of Hygiene, Timișoara, Romania
4. Centre for Studies in Preventive Medicine, "Victor Babeș" University of Medicine and Pharmacy, Timișoara, Romania
5. Academy of Medical Sciences

Abstract: The climate change induced evolution of pathogen agents, the potential of new zoonoses generating pandemics and mass circulation of people are factors that highlight the need for efficient strategies of prevention. This review paper explores the dynamic of new zoonoses, the lesson learned after the COVID-19 pandemic and the role of implementing one health concept in public health policies. By systematically reviewing a large set of studies, key aspects and recommendations for preventing future pandemics were synthesised.

The interaction between humans, animals and the environment represent a complex mechanism of infectious agent transmission. The COVID-19 pandemic has illustrated the serious consequences of uncontrolled zoonotic pathogen transmission.

Material and methods: Relevant articles were identified in scientific databases: PubMed, Web of Science. Data from 10 scientific articles was systematically extracted by analysing methodologies, advances and recommendations presented in each article.

Discussion: The factors contributing to the emergence of new zoonotic diseases are multidimensional including loss of bio-diversity, human impact on ecosystems, variations in climate, wild animal trade and the way of using and managing fields. These factors classify pathogens in distinct transmission stages, showing the risk of new pathogens that can progress from wild animal reservoirs to human transmission chains.

One health concept involves integrating human and animal health with environmental interaction in tackling new zoonotic diseases. By considering the historic context and the current implementation of the one health approach, multidisciplinary collaboration is recommended in facing emergent threats. Integrated surveillance and response systems (iSR) are defined as a collaborative response (monitorisation and coordinated acting) between human and animal health sectors in facing outbreaks with economic and epidemic advantages.

The COVID-19 pandemic emphasised the importance of understanding the provenance of pathogens, natural reservoirs and transmission patterns. The selected studies reiterate the role of immunisation, vaccination and the need for proactive prevention strategies: monitoring zoonoses, genome sequencing of new zoonotic agents at the global level, strengthening medical supply chains and vaccine production as well as developing proper legislative tools.

Early detection of new pathogens, integrative communication between sectors and bio-security measures are aspects that indicate the need for the involvement of stakeholders, the food and agriculture industry, merchants and consumers in elaborating solutions at the local level.

Conclusion: By combining various data from a set of studies, we emphasised the need to urgently approach how new zoonoses emerge. It is proved that the one health concept impacts different activity sectors, while it is pledged for integrating the animal, human and environmental surveillance systems. Having the COVID-19 pandemic as catalysing factor, a multi-disciplinary approach together with citizen involvement and elaborating evidence-based strategies are crucial in preventing future pandemics.

References

1. Miranda, A. V., Lestari, B. W., Indrarini, A., Arsy, F. F., Sagala, S., Bisri, M. B. F., & Lucero-Prisno, D. E. (2023). Adaptation of health systems to climate change-related infectious disease outbreaks in the ASEAN: Protocol for a scoping review of national and regional policies. *PLOS ONE*, 18(6), e0286869. <https://doi.org/10.1371/journal.pone.0286869>
2. Sánchez, A., Contreras, A., Corrales, J., & De La Fe, C. (2022). En el principio fue la zoonosis: One Health para combatir esta y futuras pandemias. *Informe SEESPAS 2022. Gaceta Sanitaria*, 36, S61–S67. <https://doi.org/10.1016/j.gaceta.2022.01.012>
3. Benis, A., Tamburis, O., Chronaki, C., & Moen, A. (2021). One Digital Health: a unified framework for future health ecosystems *Journal of Medical Internet Research*, 23(2), e22189. <https://doi.org/10.2196/22189>
4. Zinsstag, J., Utzinger, J., Probst-Hensch, N., Lv, S., & Zhou, X. (2020). Towards integrated surveillance-response systems for the prevention of future pandemics. *Infectious Diseases of Poverty*, 9(1). <https://doi.org/10.1186/s40249-020-00757-5>
5. Monath, T. P., Kahn, L. H., & Kaplan, B. (2010). One health perspective. *Iilar Journal*, 51(3), 193–198. <https://doi.org/10.1093/ilar.51.3.193>
6. Young, J., Coulombier, D., Domanovic, D., Zeller, H., & Gossner, C. M. (2019). One Health approach for West Nile virus surveillance in the European Union: relevance of equine data for blood safety. *Eurosurveillance*, 24(16). <https://doi.org/10.2807/1560-7917.es.2019.24.16.1800349>
7. Sridhar, D. (2022). Five ways to prepare for the next pandemic. *Nature*, 610(7933), S50. <https://doi.org/10.1038/d41586-022-03362-8>
8. Cutler, S. J., Fooks, A. R., & Van Der Poel, W. (2010). Public health threat of new, reemerging, and neglected zoonoses in the industrialized world. *Emerging Infectious Diseases*, 16(1), 1–7. <https://doi.org/10.3201/eid1601.081467>
9. Buchy, P., Buisson, Y., Cintra, O., Dwyer, D. E., Nissen, M. D., De Lejarazu, R. O., & Petersen, E. (2021). COVID-19 pandemic: lessons learned from more than a century of pandemics and current vaccine development for pandemic control. *International Journal of Infectious Diseases*, 112, 300–317. <https://doi.org/10.1016/j.ijid.2021.09.045>
10. Holmes, E. C. (2022). COVID-19—lessons for zoonotic disease. *Science*, 375(6585), 1114–1115. <https://doi.org/10.1126/science.abn2222>

Conference Abstract – L16

The Necessity Of Musculoskeletal Ultrasound In Physical Therapy Management Of Pediatric Traumatic And Non-Traumatic Injuries

Andra Pintilie, M.D., PhD Student, National Center of Pediatrival Rehabilitation “Dr. Nicolae Robănescu”

Liliana Pădure, M.D., PhD, National Center of Pediatrival Rehabilitation “Dr. Nicolae Robănescu”

Mihaela Bejan, M.D., National Center of Pediatrival Rehabilitation “Dr. Nicolae Robănescu”

Introduction

Musculoskeletal ultrasound is a non-invasive, well-tolerated and highly accessible procedure for evaluating both acute onset and chronic situations in pediatric population, from early ages of life to teenagers. The procedure allows fast evaluation and therefore the certainty diagnosis, facilitating the proper rehabilitation management and procedures.

Objective

Fast evaluation and assessment of musculoskeletal disorders, avoiding high cost and time-consuming investigations.

Material and method

Patients within the age range of 7 weeks and 16 years old, which were evaluated using two ultrasound medical devices Logiq E and portable GE Logiq E.

Sinding-Larsen-Johansson Syndrome – most frequently diagnosed in performance sportsmen teenagers (football, basketball), with repetitive patellar pressure and movement induced pain, sometimes edema, and restrictive athletic performance.

Baker cyst – most often diagnosed in male patients with age range from 4 to 8 years old, with spontaneous onset and unknown ethology.

Heterotopic calcifications – of soft tissue, in patients with long time bed ridden patients after traumatic brain injury, spinal cord injury, surgical procedures, and amputations. The bony structures developed in soft tissues are very painful and if localized in a joint, it result in functional limitation with loss of range of motion.

Encapsulated hematomas – the chronic stage of posttraumatic hematomas. Case report of a 7 week new born with obstetrical clavicular fracture.

Muscular injuries – frequently hamstring injuries in young football players, especially in femoral biceps.

Results

Using musculoskeletal ultrasound, we could rapidly identify and prescribe the proper rehabilitation management for traumatic and non-traumatic injuries.

Conclusions

Musculoskeletal ultrasound proved to be a reliable imagistic tool in fast diagnosis. Non-invasive method, with shorter procedural time in comparison with MRI, CT, also well tolerated by children.

Conference Abstract – L17

New Phenotypes In Spinal Muscular Atrophy Patients Treated With Nusinersen, Risdiplam Or Onasemnogene Abepavovec-Xioi

Authors: Mirea Andrada^{1,2}, Leanca Madalina^{1,2}, Mihaela Axente³, Grigoras Petru-Florin², Vasile Daniela Dorina²

¹University of Medicine and Pharmacy “Carol Davila”, Bucharest

²National University Hospital for Children Neurorehabilitation “Dr. Nicolae Robănescu” (NUHCN), Bucharest

³Teaching Emergency Hospital for Children “M.S.Curie”, Bucharest

Introduction. Spinal muscular atrophy (SMA) is a nervous system degenerative disorder with autosomal recessive genetic transmission, affecting motor neurons in the anterior medullary horn, often those in the brainstem and leading to their death with loss of muscle mass and motor deficiency. SMA is caused by deletion or mutation of the survival motor neuron 1 (SMN1) gene, and the nearly identical SMN2 gene fails to generate adequate levels of functional SMN protein due to a splicing defect. Currently, three therapeutics targeted to increase SMN protein are available in Romania: Onasemnogene Abeparvoce, Nusinersen and Risdiplam settled by the National Health Program. The best results have been obtained in presymptomatic patients. Few fields of medicine have seen such rapid expansion of the therapeutic options and the magnitudes of clinical response as those seen in the field of SMA. These new therapies are providing us with new information about the biology and pathogenesis of SMA, including the timing of its initiation, the steps that lead to progressive muscle weakness and the possibility that the pathological process can be reversed or attenuated, especially with early intervention.

Objectives. Clinical responses in patients with symptomatic SMA can reach levels that were unanticipated, but the longer the disease duration and the greater the severity, the more modest is the response. Aware of the need to apply the standards of care in this disease, we have designed this presentation in order to demonstrate the high efficiency and good motor achievements depend on the age/disease stage at which therapy is initiated.

Material and methods. The paper aims to reproduce images with patients, methods of diagnosis and innovative treatments used in NCHCN. Current options are effective in improving mobility, good ventilation and improvements in ventilation free survival in the patient who have started early treatment.

Results. We have observed disease trajectories that differ significantly from the known natural history of the disease. These new phenotypes now also cross the traditional subtypes of SMA. It is now more appropriate to rely on a combination of age of onset, number of SMN2 copies, and age at start of drug treatment rather than the traditional subtypes to define a clinical phenotype of SMA.

Conclusions. The advances in therapy for SMA have improved survival and quality of life, and this poses new challenges. An early treatment leads to a better result and we need to improve our diagnostic ability and reduce all the procedures in order to ensure a fast treatment. We need to guarantee the best standards of care to get the best results and to describe new phenotypes in SMA patients. We need consensus on SMA-type classification and endpoints that determine intervention efficacy of any treatment. Standard newborn screening seems to be an appropriate tool to achieve maximum treatment effects, a timely diagnosis and treatment initiation.

Conference Abstract – L18

Rare Causes Of Congenital Cataracts In Children

Andra Pintilie¹, Elena-Silvia Shelby¹, Liliana Padure^{1,2}, Mihaela Axente^{1,3}, Mihaela Bejan¹, Mihaela Badina^{1,3}, Elena Neagu¹, Corina Sporea^{1,2}, Andrada Mirea^{1,2}

1. „Dr. Nicolae Robanescu” National Clinical Center for Children’s Neurorehabilitation, Bucharest
2. Chair of Balneofiziokinetotherapy and Rehabilitation, Faculty of Midwifery and Nursing, „Carol Davila” University of Medicine and Pharmacy, Bucharest
3. Chair of Pathophysiology II, Faculty of General Medicine, „Carol Davila” University of Medicine and Pharmacy, Bucharest

Congenital cataracts – the clouding of the lens of the eye seen at birth or in the first year of life, can have multiple causes, among which certain prenatal infections (rubella, syphilis, CMV), genetic mutations (CRYAA, CRYBB1, CRYBB2, CXN) or chromosomal aberrations. One of the less-known syndromes associated with congenital cataracts is CCFDN – congenital cataracts, facial dysmorphism – neuropathy syndrome. According to various sources of the literature there are less than 200 patients ever reported to have this syndrome, which is caused by a founder mutation and present only in the Roma population, due to endogamy. Here we present a 10-year old girl with congenital cataracts, facial dysmorphism specific to this syndrome and sensory and motor neuropathy; the patient was admitted to Dr Nicolae Robanescu Children’s Hospital for medical rehabilitation; genetic testing confirmed our suspicion of CCFDN syndrome revealing the founder mutation present in all patients diagnosed so far. We believe presenting this case is important for three reasons, namely: 1. patients with CCFDN syndrome are prone to infectious rhabdomyolysis and life-threatening complications during anesthesia, in the case of major surgeries – careful monitoring and the avoidance of certain anesthetics are mandatory; 2. to our knowledge, in spite of the fact that the prevalence of CCFDN is 1,4% in the Roma ethnicity and currently there are 569.000 Roma living in our country, our case is only the 4th to ever be reported in Romania, suggesting the need to disseminate information that can make this syndrome more easily recognizable; 3. the high prevalence of this disease in the Roma population due to the prevailing founder mutation, as well as of other recessive diseases caused by founder mutations suggests the urgent and massive need of a counselling program amongst the Roma population that should advise against endogamy.

Conference Abstract – L24

Newborn Screening For Spinal Muscular Atrophy: Early Detection For Improved Outcomes

Authors: Mirea Andrada^{1,2}, Leanca Madalina^{1,2}, Dima Vlad³, Shelby Elena Silvia², Neagu Elena²

¹University of Medicine and Pharmacy "Carol Davila", Bucharest

²National University Hospital for Children Neurorehabilitation "Dr. Nicolae Robănescu" (NUHCN), Bucharest

³Clinical Hospital "Filantropia", Bucharest

Background: Spinal Muscular Atrophy (SMA) is a rare genetic disorder that affects the motor neurons in the spinal cord, leading to muscle weakness and atrophy. It is the leading genetic cause of infant mortality, with symptoms appearing in the first few months of life. Early diagnosis and intervention are crucial for improving outcomes and providing appropriate care for affected infants. Newborn screening (NBS) has emerged as a powerful tool in identifying infants with SMA before symptoms manifest.

Discussions: Newborn screening for SMA involves testing a small blood sample taken from a newborn's heel. This sample is then analyzed for the presence of specific genetic markers associated with SMA. The introduction of NBS for SMA has revolutionized the diagnosis and management of this condition. It allows for early identification of affected infants, enabling prompt medical intervention and access to specialized care. Early diagnosis through NBS offers several advantages. Firstly, it allows for timely initiation of treatment, such as gene therapy or other disease-modifying therapies, which have shown promising results in halting or slowing the progression of SMA. Secondly, it provides an opportunity for genetic counseling and family planning, allowing parents to make informed decisions about future pregnancies. Additionally, NBS helps identify carriers of the SMA gene, enabling them to seek appropriate genetic counseling and testing.

Conclusion: In conclusion, newborn screening for spinal muscular atrophy is of utmost importance. It plays a vital role in early detection, intervention, and management of this devastating genetic disorder. By identifying affected infants before symptoms appear, NBS allows for timely treatment initiation, improving outcomes and quality of life. Furthermore, it facilitates genetic counseling and family planning, empowering parents to make informed decisions. The implementation of NBS for SMA has the potential to save lives, reduce the burden on affected families, and contribute to ongoing research and advancements in the field. It is crucial that healthcare systems worldwide recognize the significance of newborn screening for SMA and work towards its widespread implementation.

Conference Abstract – L25

Functional Electrical Stimulation In Pediatric Patients. Our Experience In Applying Bioness Devices

Author: Constantin Florin Dragan

In the Dr. Nicolae Robanescu National Clinical Neuropsychomotor Recovery Center for Children, these modern Bioness devices have been applied to pediatric patients since 2018. We have at our disposal the complete set of functional electrical stimulation both for the pelvic limbs (calf, both sizes and thigh) and for the thoracic limbs, both sizes, for children and for adolescents with adult sizes. The L 300 Go system is indicated to produce dorsiflexion of the ankle in cases of droop foot, flexion/extension of the knee or flexion/extension of the fist and fingers for the thoracic limb, in the case of the H200 device, stimulating weakened muscles, in conditions of central motor neuron syndrome, cerebral palsy, craniocerebral trauma, spinal cord pathologies, multiple sclerosis. It is very important to know and respect the indications and contraindications of the Bioness device. It is recommended for children over 5-6 years old, with one of the previously mentioned conditions and who cannot actively perform the movements mentioned above.

Key words: cerebral palsy, spasticity, droop foot, functional electrical stimulation, L300GO, H200

Conference Abstract – L26

Chronic Progressive Gait Disorder With Prepubescent Onset Epiphysiolysis

Madalina Leanca¹, Diana Carabageac^{1,2}, Andreea Suciuc^{1,2}, Nicoleta Chiriloi

¹ National University Hospital for Children Neurorehabilitation "Dr. Nicolae Robănescu" (NUHCN)", Bucharest, Romania

²"Carol Davila" University of Medicine and Pharmacy, Neurosciences Department, Pediatric Neurology Discipline, Bucharest, Romania

Objectives: Femoral epiphysiolysis is a condition of the cervico-cephalic conjugation cartilage that no longer firmly fixes the femoral head on the neck, allowing it to tilt. The evidence and ongoing studies evaluate the triggering causes, the risk factors associated with this pathology and the orthopedic and surgical treatment, as well as the appropriate physical-kinetic recuperative program.

Methods: We report a 14-year-old male patient, with normal intellect, overweight, who was diagnosed with right femoral epiphysiolysis 4 years ago, initially presenting chronic progressive pain in the coxofemoral joint, following the surgical indication. The patient has compensatory left dorso-lumbar scoliosis and dorsal kyphosis degree 2. He underwent surgical treatment: modified Dunn method operation, fixation with an external fixator and definitive epiphysiodesis distal to the left femur, orthopedic treatment: wearing orthopedic shoes with elevation on the right side and physical-kinetic treatment.

Results: After the surgical treatment, where the right coxofemoral joint was reduced and fixed in situ, followed by the physical-kinetic recovery program (exercises to increase muscle strength, pain re-education, muscle toning exercises and gait correction including robotic recovery) the improvement of walking and the amelioration of algodysfunctional syndrome were observed.

Conclusions: In all degrees, the treatment is surgical, the technique used depends on the degree, form and stage of the disease. It is important for the patient to present himself within the first 3 weeks from the onset in order to considerably increase the success of the treatment and to reduce the risk of further complications. Physical-kinetic treatment is essential for a favorable evolution. For a good prognosis and to avoid large-scale operations with unpredictable results, it is essential to establish a correct and prompt diagnosis and treatment.

Conference Abstract – L28

Implications Of Heart Injury In Duchenne Muscular Dystrophy

Amelia Aria¹, Cristina Filip¹, Bianca Petrescu¹, Mădălina Vulcan¹, Georgiana Nicolae²

1. MS Curie Emergency Children Hospital, Bucharest

2. CNRNC N Robanescu Children Rehabilitation Hospital, Bucharest

Duchenne muscular dystrophy (DMD) is an x-linked genetic neuromuscular disorder that occurs in 1 in 5,000 male subjects due to mutation of the DMD isoform gene Dp427m, leading to extreme vulnerability of sarcolemma.

The clinical picture is varied, muscular and cardiac symptoms being the most important complications. Those complications ultimately leading to cardiac and respiratory failure that are the main cause of mortality and morbidity after the second decade of life.

Cardiac involvement is expressed by symptoms of heart failure, chest pain, ECG and echocardiographic changes.

The objective of periodic cardiac monitoring is to establish the moment of initiation of cardiological treatment to preserve cardiac function and reduce the symptoms of heart failure.

The new echocardiography techniques (speckle tracking and strain rate) early detect the alteration of the contractility of the left ventricle. The gold standard technique for monitoring cardiac function is represented

by MRI (late gadolinium uptake), but in its absence, echocardiographic and ECG evaluations obtain comparable results.

DMD is a serious neuromuscular disease with a poor prognosis; dilated cardiomyopathy, arrhythmias and heart failure represent the main cause of morbidity and mortality in the pediatric cohort. Early diagnosis of the typical changes for dilated cardiomyopathy and early treatment with converting enzyme inhibitors, sartans, beta-blockers remained the basic interventions that improve the prognosis.

Conference Abstract – L29

The Therapeutic Importance Of The Posture Of The Patient With Spinal Amyotrophia

Mihail-Alexandru Dumangiu^{1,2}, Tiberiu Tătaru³

¹National Clinical Center for Children's Neurorehabilitation „Dr. Nicolae Robănescu” Bucharest

²Spiru Haret University, Bucharest, Romania

³Constantin Brâncuși University, Târgu Jiu, Romania

Abstract:

Introduction: Spinal muscular atrophy (SMA) is part of a group of autosomal recessive degenerative diseases that affect the motor alpha neurons in the spinal cord, the consequences being progressive muscular atrophy, weakness and paralysis. In 2007 a report of the consensus declaration makes it possible for the guidelines that underpin the care guides and protocols to be adopted at the international level. Children with spinal muscular atrophy face difficulties lifting the head off the supporting surface or maintaining the head upright when lifted into seated position, deficit that progresses and spreads to the distal extremity of the limbs.

Method: Muscle weakness and the positions in which children with SMA are held, determines in time disorders of vertebral statics, deformation of the thoracic cage, associated with respiratory deficiency, muscle retractions in the upper and lower limbs, and at the neck muscles level, but also fascia and capsulo-ligamentous retraction. Posturing implies an attitude imposed on the whole body or at least on parts of it, for therapeutic or preventive purposes, to correct or avoid the installation of static deviations and vicious positions or to facilitate a physiological process. The duration of the posturing is variable, depending on the specific characteristics of each patient and the purpose it is made for. Patients with spinal muscular atrophy with a higher motor deficit, remain in certain positions for a longer period of time. These positions are not always the most accurate. The fact that they are placed in different positions on the bed for rest, in carts during the day, or they are held in an uncontrolled manner, makes these positions become true posturings. For the prevention or rehabilitation of deformations, have been used several medical devices, such as pillows, sandbags, corsets, orthoses and other devices. The physical therapy program includes manual orthopedic therapy, hydrotherapy, ergotherapy and robotic therapy.

Conclusion: Early implementation of posturing and physical therapy help maintaining the joints functional, muscles preserve their characteristics, breathing does not become a risk factor.

Key words: Spinal muscular atrophy (SMA), posture, rehabilitation, early intervention.

Conference Abstract – L31

Human Gait Prediction Using Machine Learning Techniques

Diana MARUSIC¹, Galina MARUSIC², Gelu ONOSE^{3,4}, Andrei BRAGARENCO²

¹ Information Systems and Machine Learning Lab, University of Hildesheim, Germany

² Department of Computer Science and Systems Engineering, Technical University of Moldova, Republic of Moldova,

³ Department of Physical and Rehabilitation Medicine, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania,

⁴ Emergency Clinical Hospital Bagdasar Arseni, Bucharest, Romania

Abstract: This paper addresses the challenge of predicting human walking kinetics through the utilization of Artificial Intelligence (AI) models. It presents a study in the field of Smart Gait, which includes integrated systems for human gait analysis using AI techniques. The study examines the aspects of integrating AI models with a robotic exoskeleton, aimed at facilitating the mobility of individuals with limited mobility. A compelling case study is presented, demonstrating the practical application of an AI model to address the formulated issue.

Keywords: Artificial Intelligence (AI), Smart Gait, AI model, exoskeleton.

Conference Abstract – L32

Architectural Approach Of Sensor Actuator Abstraction For A Biped Exoskeleton

Andrei BRAGARENCO¹, Galina MARUSIC¹, Gelu ONOSE^{2,3}, Diana MARUSIC⁴

¹ Department of Computer Science and Systems Engineering, Technical University of Moldova, Republic of Moldova

² Department of Physical and Rehabilitation Medicine, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania,

³ Emergency Clinical Hospital Bagdasar Arseni, Bucharest, Romania,

⁴ Information Systems and Machine Learning Lab, University of Hildesheim, Germany

Abstract — The paper comes with an architectural approach in sensor-actuator interactions for an exoskeleton of the lower limbs. The functionalities of the system components that implement the interaction with the external environment, such as sensors and actuators, are implemented with a complex combination of technologies from the fields of mechanical, electrical and software engineering. In order to control this mix of technologies, a special architectural approach is needed that would take into account this variety of technologies, abstract from them and provide the upper levels of the system with interfaces for interaction with the environment. Such a mix of interaction technologies is also specific for an exoskeleton of the lower limbs.

Keywords — Layered Architecture, Sensor, Actuator, Exoschelet

Conference Abstract – L33

AI- Based Solutions In Neuro-Investigations

Nirvana Popescu*, Asma Channa*, Rares Ifrim*, Aura Loredana Popescu*

*University POLITEHNICA of Bucharest, Computer Science Department

Background. In recent years, in the context of medical neuro-investigations, the solutions based on artificial intelligence offer robust automatic evaluations that can help in determining effective medical decisions. The present work addresses two aspects that determined the development and experimental testing of two solutions that deal with conditions that appear at the extreme ages of patients, more precisely the evaluation of the emotional state of children diagnosed with autism and the evaluation of the evolution of Parkinson's disease in the elderly, based on machine learning techniques and wearable devices. Autism represents a neurodevelopmental disorder that is characterized by problems in expressing feelings, integrating socially, and having repetitive behaviors. Regarding to the second topic, Parkinson's Disease (PD) is one of the prominent neuro-cognitive disorder (NCD), which typically appears after the age of 60.

Methods. Drawings are an important part of a child's life and drawing interpretation is very useful to find out what the child is feeling. In this context, this work describes an android application called PandaSays that was developed based on the drawing's interpretation, as an improved eHealth system dedicated to autism, to help parents and tutors communicate better with their children and to understand their emotional state. The algorithm used in the android application interprets the children's drawings and identifies their affective state. Recent studies tried to predict the affective state of the child using humanoid robots or using images of facial expressions. There is no application that uses drawings to evaluate the affective state of a child. By means of this solution, drawings will become a gate to the child's affective state and behavior. Another important aspect of the application is that the parent does not have to go to a psychologist to interpret his/her child's drawings, the application does that for him, as the dataset is already validated by a certified psychologist. The solution incorporates a machine learning algorithm that detects the affective state of the child ("happy", "angry", "sad", "insecure", and "fear") from what he/she draws. The application's dataset contains 1453 drawings. The output is sent further to one of the robots: Marty, Alpha1P or Alpha 1E. The robots will execute a specific action, depending on the state previously received from the machine learning algorithm.

For the solution dedicated to PD state evaluation, a novel framework is developed for assessment of motor deficits from upper extremities of NCD patients. The suggested system is based on wearable technology and cloud computing. The proposed approach is an integration of signal processing, resampling methods and deep learning classifiers. The recommended system addresses numerous challenges by developing a small form-factor wearable bracelet named A-WEAR which provides movement related data continuously connected with smart phone using BLE, mobile application as user interface, a network that enables wirelessly to MS Azure cloud, which provides the ServiceNow platform as a web application and builds a database utilizing the movement information from the A-WEAR.

Results. PandaSays mobile application dedicated to the affective state evaluation of children conforms to the accessibility guidelines. A robust set of experiments has been done in order to validate the proposed innovative solution, the accuracy being of 98% and the children's response to the robots' interaction exceeding our expectations.

In the case of PD evaluation, in order to avoid classifier bias because of imbalance data distribution, resampling is performed which consequently improved tremor severity estimation with accuracy 96%, IBA=97%, F1-score=97%, G-mean=97% and AUC=99%. Randomly sampling worked better than under and hybrid sampling. The XGBoost and CatBoost classifiers provide the best performance to evaluate tremor severity while patients are on OFF and ON state (with or without medication), according to the performance comparison of several classifiers.

Thus, the proposed evaluation solutions are based on various hybrid machine learning techniques in combination with wearable devices, bringing innovative solutions to health problems.