

Turin, 08 - 10 July 2024





Book of Abstracts



Turin, 08 - 10 July 2024





Managing Uncertainty in Artificial Intelligence: Assessment, Modeling, Decision-Making

Robertas Alzbutas^{1,2}

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Short bio of speaker:

Professor Dr. Robertas Alzbutas is a Leading Researcher in the Research Group of Al, Data Analytics, and Modelling at Kaunas University of Technology and a Senior Research Associate at the Lithuanian Energy Institute, with a PhD in Technological Sciences since 2004. He has extensive expertise across Data Science, Mathematics, Al, IT, Medicine, Econometrics, and Civil Engineering, including Safety Engineering. His research focuses on rare data processing, risk assessment, Bayesian inference, big data analytics, machine learning, and probabilistic safety assessment. He has led various national and international projects, supervised students and researchers at all levels, and co-authored over 100 highly cited publications.

Abstract:

As artificial intelligence (AI) continues to spread in various domains, understanding and managing uncertainty are crucial for robust and reliable AI systems. This lecture explores the wide landscape of uncertainty and AI, focusing on key aspects such as uncertainty assessment, modelling techniques, and effective management strategies. The topics covered include: (1) modelling data/results uncertainty and employing uncertainty measures; (2) distinguishing between local and global uncertainty estimates through sensitivity analysis; (3) establishing tolerance intervals to handle computationally intensive calculations; (4) application of Monte Carlo and sampling methods for black-box models; (5) assessing the sensitivity of uncertainty using variance-based methods; (6) integrating uncertainty into the decision-making process for practical outcomes; and (7) evaluating precision and accuracy measures with illustrative examples. By going into these areas, this lecture aims to provide practitioners and researchers with essential insights and tools to navigate and mitigate uncertainty in AI applications effectively.



Turin, 08 - 10 July 2024





Enhancing Detection of Brain Injuries with Deep Learning and Uncertainty Analysis

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- 2. Lithuanian Energy Institute; Breslaujos str. 3, Kaunas 44403, Lithuania; robertas.alzbutas@lei.lt

Short bio of speakers:

Professor Dr. Robertas Alzbutas is a Leading Researcher at KTU with a PhD in Technological Sciences since 2004. His research focuses on rare data processing, risk assessment, Bayesian inference, big data analytics, machine learning, and probabilistic safety assessment. He has led various projects, supervised students at all levels, and co-authored over 100 publications.

Jewel Sengupta is currently pursuing a PhD in Informatics Engineering at KTU. He also served as a Project Junior Researcher from 2023 focusing on innovative technology for predicting and providing early warnings of delayed cerebral ischemia after subarachnoid hemorrhage. He is a dynamic and accomplished professional with a passion for technology and AI.

Abstract:

Brain injuries, including subarachnoid hemorrhage (SAH) and other types of incranial bleeding, pose significant challenges in acute stroke management and contribute substantially to disability-adjusted life years lost. Early detection and treatment are crucial. This study explores the use of deep learning (DL) techniques for improving the detection and monitoring of brain injuries using non-contrast computed tomography (NCCT) head imaging.

Our approach involves modifying conventional segmentation models by integrating both segmentation and regression tasks, enhancing the localization and quantification of SAH. A novel region-growing method segments brain regions affected by SAH. Features are extracted from pre-trained models, and dimensionality reduction is performed using optimization algorithms. An unsupervised DL method for automatic segmentation of SAH, based on spatial distance, provides regular contours with short processing times. Optimized feature vectors are used in an unsupervised classification model to classify SAH subtypes.

Incorporating uncertainty analysis is a key component of our framework. Despite challenges such as limited labeled datasets and the complexity of volumetric image analysis, our results demonstrate significant improvements in diagnostic accuracy and predictive capabilities. The incorporation of DL algorithms in detecting brain injuries using NCCT imaging represents a significant advancement in acute care diagnosis and management. Precision and accuracy are essential metrics for evaluating these algorithms' performance, directly impacting clinical decisions and patient outcomes. This comprehensive approach facilitates validation, estimation of prediction performance, and effective uncertainty reduction, aiding clinicians in making well-informed decisions for better patient outcomes and more effective management of brain injuries.



Turin, 08 - 10 July 2024





Hammersmith Infant Neurological Examination (HINE) and General Movements in infants for the prediction of neurodevelopmental outcomes

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Short bio of speaker(s):

Umut Apaydin PT, PhD: He completed his master of science (2016) and PhD (2021) in the Department of Physical Therapy and Rehabilitation at the Gazi University. He has focused on early intervention and early diagnosis of infants at risk. Now, he is working at Karadeniz Technical University as an Assistant Professor.

Turgay Altunalan PT, PhD: He graduated with an MSc degree from Istanbul University in 2007 and a Ph.D. degree in 2021 from Marmara University. He has focused on early intervention and early diagnosis of at-risk babies. He worked in a nonprofit organization called Cerebral Palsy Turkey between 2010 and 2022. Now, he is working at Karadeniz Technical University as an Assistant Professor.

Abstract:

Hammersmith Infant Neurological Examination (HINE) and Genaral Movements (GMs) are widely use assessment tool for prediction neurodevelopmental disabilities like Cerebral Palsy (CP). The predictive power of the HINE test battery performed at 3 months was 88% sensitive and 62% specific (Morgan et al., 2019). Similarly, sensitivity and specificity rates of the HINE test battery performed at 3-6 months in children with hypoxic ischemic encephalopathy (HIE) who received cooling therapy were reported as 83.3% and 87.8%, respectively (Apaydin et al., 2021). In another study, infants with HINE scores of ≤56 at 3 months and ≤65 at 12 months showed high sensitivity and specificity (~90%) for the diagnosis of CP. In a study, it was stated that the sensitivity and specificity ratio of GMs assessment performed during the fidgety period to predict CP was 95% and 97%, respectively (Morgan et al., 2019). According to the study of Prechtl et al., (1997) the specificity was 96% and sensitivity was 95% in predicting CP (Prechtl et al., 1997). In a study conducted in infants with HIE who received cooling therapy, the rate of prediction of CP by GMs was reported as 83.3% sensitivity and 100% specificity (Apaydin et al., 2021). Considering the results of the literature, it is necessary to use the GMs evaluation method in terms of its early predictive power for the diagnosis of neurodevelopmental delays. Thus, our presentation will describe HINE and GMs. The literature on HINE and GMs will be summarized. Also, artificial intelligence in GMs will be described.



Turin, 08 - 10 July 2024





Hands-on: AI for medical imaging predictions

Mateusz Bednarski¹

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Short bio of speaker(s):

Experienced software engineer, working on large-scale web applications for hospitals in the USA. Previously involved in developing disease incidence prediction models for University Hospital in Krakow, Department of Neonatology. Currently focused on exploring machine learning models' development cycle automation, integrating machine learning models in web applications and architecting ML-enabled cloud infrastructures.

Abstract:

This workshop will be a guide on how to build an image classifier, based on the convolutional neural network (CNN). We'll work on the APTOS 2019 – Blindness Detection dataset, consisting of retina images taken using fundus photography. We'll attempt to build a classifier detecting diabetic retinopathy. We'll start by introducing the specifics of working with images and ways of preparing them to be later used as a ML models' input. During the workshop we'll test different architectures of CNNs and compare their results. The workshop will be conducted, based on a Google Colaboratory notebook. Every concept will be first introduced with code examples explained by the trainer, and then complemented with a set of self-paced tasks. After the workshop you will be able to train a CNN-based classifier for the use on your image set.



Turin, 08 - 10 July 2024





Hands-on: Implementation of shallow ML techniques

Mateusz Bednarski¹

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Short bio of speaker(s):

Experienced software engineer, working on large-scale web applications for hospitals in the USA. Previously involved in developing disease incidence prediction models for University Hospital in Krakow, Department of Neonatology. Currently focused on exploring machine learning models' development cycle automation, integrating machine learning models in web applications and architecting ML-enabled cloud infrastructures.

Abstract:

This workshop will be a guide on how to build a classifier, based on the shallow machine learning techniques (Logistic Regression and Random Forest). We'll follow the complete process, similar to the one that you may face in the future, while working on your dataset. We'll start by importing the dataset, consisting of physical parameters of a fine needle aspirate (FNA) of a breast mass. We'll evaluate the dataset's statistics and prepare it (by reshaping and normalization) to be used for the training of the machine learning methods. We'll examine the quality of the models by calculating the metrics, based on their predictions. We'll tackle the problem of possible overfitting and ways of avoiding it while learning. The workshop will be conducted, based on a Google Collaboratory notebook. Every concept will be first introduced with code examples explained by the trainer, and then complemented with a set of self-paced tasks. After the workshop you will be able to train a shallow-ML-model-based classifier by your own.



Turin, 08 - 10 July 2024





Hands-on: Practical data handling and processing for ML

Mateusz Bednarski¹

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Short bio of speaker(s):

Experienced software engineer, working on large-scale web applications for hospitals in the USA. Previously involved in developing disease incidence prediction models for University Hospital in Krakow, Department of Neonatology. Currently focused on exploring machine learning models' development cycle automation, integrating machine learning models in web applications and architecting ML-enabled cloud infrastructures.

Abstract:

During the workshops we'll work on developing basic understanding of how to work with data in Python programming language, to later be able to transform them to a format required by the selected machine learning method. We'll start with the introduction of the dataframe data structure, present in many data-processing frameworks, and ways of importing our dataset to it. Then we'll explore different approaches to querying the data and performing aggregations. We'll also learn how to get some statistical insights about the dataset. The workshop will be conducted, based on a Google Colaboratory notebook. Every concept will be first introduced with code examples explained by the trainer, and then complemented with a set of self-paced tasks. After the workshop you will be able to import, reshape and manipulate your data with Python.



Turin, 08 - 10 July 2024





EEG in Pediatric Neurology: Current Practices and Emerging Challenges

Daniele Marcotulli¹, Carlotta Canavese¹

1. Pediatric Neurology Unit, Ospedale Infantile Regina Margherita, Torino. <u>daniele.marcotulli@unito.it</u>, <u>ccanavese@cittadellasalute.to.it</u>

Short bio of speaker(s):

Daniele Marcotulli: I am a child and adolescent neuropsychiatrist working at the University Hospital of Turin, where I concluded my residency program. I obtained a PhD at the Polytechnic University of Marche (Italy), while I conducted part of my Ph.D. project as a research fellow in the Bajjalieh Lab at University of Washington (Seattle). I have a keen interest in brain physiology and pathological mechanisms and have cultivated this interest during the PhD years, when I had the opportunity to study the basic neurobiological mechanisms of epilepsies in mice.

Carlotta Canavese: I am a child and adolescent neuropsychiatrist working at the University Hospital of Turin, where I am the director of the Pediatric Neurology Unit. My main clinical interests include electrophysiology, epilepsies and movement disorders.

Abstract:

Electroencephalography (EEG) is the most widely utilized diagnostic tool in child and adolescent neurology, offering invaluable insights into brain maturation, physiology, and pathological processes. EEG recordings are based on unipolar or bipolar electrical potential differences generated by the propagation of action potentials in the brain cortex. These recordings provide excellent temporal resolution and variable spatial resolution.

The features analyzed in the clinical interpretation of EEG recordings include frequency, amplitude, spatial characteristics of the traces, their variations across different scalp regions, and the presence or absence of specific physiological and pathological graphoelements.

The physiological features captured by EEG undergo significant changes throughout development, with the most pronounced transformations occurring within the first three years of life. These developmental changes are further influenced by various behavioral states, such as wakefulness and different sleep stages.

In addition to monitoring brain development, EEG is crucial in diagnosing several pathological conditions, including epilepsies, toxic, ischemic and metabolic encephalopathies, and inflammatory processes. Despite its long history, having been invented nearly a century ago, EEG interpretation has predominantly relied on clinical expertise and the visual detection of physiological and pathological features. Although there have been notable efforts to create automated EEG interpretation systems, these systems have struggled to effectively address the complexities inherent in the developing brain.



Turin, 08 - 10 July 2024



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Exploring the pathogenesis of neurodevelopmental defects: the NeuroWES project

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1 Department of Pediatrics and Public Health and Pediatric Sciences, University of Torino, Italy; 2 Molecular Biotechnology Center "Guido Tarone University of Torino, Italy; 3 Department of Medical Sciences, University of Torino, Italy; 4 Department of Molecular Medicine, University of Pavia, Italy; 5 IRCCS Mondino Foundation, Pavia, Italy; 6 Department of Oncology and Molecular Medicine, Istituto Superiore di Sanità, Roma, Italy; 7 Medical Genetics Unit, Città della Salute e della Scienza University Hospital, Torino, Italy; 8 Hemoglobinopathy Reference Center, San Luigi Gonzaga University Hospital, Orbassano, Italy; 9 Department of Sciences for Health Promotion and Mother and Child Care, University of Palermo, Palermo, Italy; 10 Medical Genetics Unit, Sant'Orsola-Malpighi University Hospital, University of Bologna, Bologna, Italy; 11 Molecular Genetics and Functional Genomics, Ospedale Pediatrico Bambino Gesù IRCSS, Roma, Italy; 12 Seaver Autism Center, Icahn School of Medicine at Mount Sinai, New York, USA; 13 Department of Clinical and Biological Sciences, University of Torino, Italy giovannibattista.ferrero@unito.it

Short bio of speaker(s): Prof. Ferrero is a Pediatrician and a Clinical Geneticist committed to the clinical and molecular characterization of rare diseases. During the doctoral training he contributed to the discovery of several disease genes, and afterwards he developed a comprehensive approach to pediatric complex diseases, describing new molecular mechanisms, genotype/phenotype correlations, and specific follow-up strategies for rare disease. Subsequently he has investigated the genetics of neurodevelopmental defects using a comprehensive genomic approach based on Array-CGH, Whole Exome Sequencing and Epigenetic Analyses. Recently he has been appointed as Head of the Hemoglobinopathies Reference Center at the San Luigi Gonzaga University Hospital, where he coordinates several Clinical studies exploring new therapeutic approach for these common mendelian disorders. The intersection between genetics, genomics and clinical care of patients affected by rare genetic diseases can be considered the focus of his entire career.

Abstract:

Neurodevelopmental disorders (NDD), which include autism spectrum disorders (ASD) and intellectual disability (ID), represent a major health issue, whose genetic bases are not yet fully delineated. We selected 467 probands with NDD (142F/325M) for family-based exome sequencing (ES) in collaboration with the Autism Sequencing Consortium coordinated by the Icahn School of Medicine at Mount Sinai (New York, USA). Sequencing and data analysis were completed for 392 families. We found likely pathogenic/pathogenic (LP/P) variants in 135 cases (34.4%). In the sub-cohort of patients with ID without ASD the diagnostic rate was 46.3% (95/205), in the patients with ASD without ID 11.2% (10/89), and in the patients presenting ID associated with ASD 30.6% (30/98). Moreover we identified 30 potentially pathogenetic variants in new candidate genes, currently under study to confirm their role in NDD and 2 variants in already known genes for which we hypothesize a new associated phenotype.

In conclusion, our data confirm that a comprehensive genomic analysis including ES has a high diagnostic rate in NDD. In our survey NDD cases with pathogenic variants in known disease genes are characterized by a wider phenotypic spectrum than expected, ranging from complex syndromes associated with ASD and/or ID to isolated ASD. We further show the importance of an unbiased family-based ES strategy for genetic diagnosis based on a periodical re-evaluation of sequencing data and deep phenotyping and clinical follow-up of patients.



Turin, 08 - 10 July 2024





From research to cotside care: a pathway of translation for AI algorithms in neonatal care

John M. O'Toole¹

1. CergenX, Dublin, Ireland. jotoole@ucc.ie

Short bio of speaker(s):

John O'Toole is Head of AI Research at CergenX, a spin-out company from the INFANT Research Centre, University College Cork, Ireland. John is working with the team at CergenX to develop robust AI models for early detection of brain injury in newborns. He is a former Research Fellow and Investigator at the INFANT Research Centre.

Abstract:

EEG is the gold-standard for seizure surveillance and detection of encephalopathy in the neonatal intensive care unit (NICU). Despite its clinical utility, the use of continuous EEG is limited by the need for expert interpretation, which is not always available. Automated methods to detect seizures and grade encephalopathy are being developed to address this limitation. Yet there has been little translation of these research methods to the cotside. CergenX, a spin-out company from the INFANT Research Centre, is developing a cotside device for automated detection of neonatal encephalopathy. This presentation will describe the many opportunities and challenges in translating AI research to cotside care, using the development of the CergenX Wave device as an example.



Turin, 08 - 10 July 2024





Introduction to shallow and deep AI techniques

Pawel Kulakowski¹

1. Institute of Telecommunications, AGH University of Krakow (kulakowski@agh.edu.pl)

Short bio of the speaker:

Pawel Kulakowski, PhD, is an associate professor in the Institute of Telecommunications at AGH University of Krakow. He also provides training on AI and machine learning for both IT companies (Ericsson, Blare) and medical societies (jENS conference, AI4NICU events). He is/was involved in European research projects, serving in Management Committees of COST Actions: IC1004, CA15104 IRACON, CA20120 INTERACT, and CA20124 AI4NICU, focusing on topics of nano-networking, AI in neonatology, wireless sensor networks, indoor localization and wireless communications in general. He was recognized with several scientific distinctions, including three awards for his conference papers and a scholarship for young outstanding researchers.

Abstract:

This talk will be a brief introduction to the whole artificial intelligence area. Different AI domains will be presented focusing on the machine learning approach. A distinction between shallow and deep techniques will be given and typical tasks like regression and classification will be discussed. Then, the process of learning on the basis of provided data will be introduced. The notions like the algorithm hypothesis, cost function, gradient descent and learning rate will be described. The over-fitting (high variance) and under-fitting (high bias) scenarios will be shown. Selected algorithms, namely linear/logistic regression and random forests will be explained in detail.



Turin, 08 - 10 July 2024





Introduction to AI for medical data predictions

Pawel Kulakowski¹

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Short bio of the speaker:

Pawel Kulakowski, PhD, is an associate professor in the Institute of Telecommunications at AGH University of Krakow. He also provides training on AI and machine learning for both IT companies (Ericsson, Blare) and medical societies (jENS conference, AI4NICU events). He is/was involved in European research projects, serving in Management Committees of COST Actions: IC1004, CA15104 IRACON, CA20120 INTERACT, and CA20124 AI4NICU, focusing on topics of nano-networking, AI in neonatology, wireless sensor networks, indoor localization and wireless communications in general. He was recognized with several scientific distinctions, including three awards for his conference papers and a scholarship for young outstanding researchers.

Abstract:

This will be a continuation of the topic of machine learning algorithms, this day focusing on deep learning approaches, with the applications for medical data. First, the motivation for the development of deep learning will be given, which started the last 'AI summer' about a decade ago. Then, artificial neural networks will be introduced, beginning from a model of a single neuron, then discussing the networks' architectures and the learning process (back-propagation). The parameters like epochs, batch size, optimizers and activation functions will be introduced in the context of hyper-parameter tuning. Then, a case with skewed (not balanced) classes will be considered. The architecture of convolutional neural networks, being the most common solution for signal and images recognition, will be presented. Finally some examples, like seizure detection on EEG data, will be provided.



Turin, 08 - 10 July 2024





Edge-AI for neonatal monitoring in NICUs

Lionel C Gontard¹

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Short bio of speaker(s):

Lionel Cervera Gontard holds a BSc in Physics from the University of Seville, Spain, an MSc in Microelectronics from IMSE-CNM, Spain, and a PhD in Materials Science from the University of Cambridge, UK. He has worked in technology companies and in research centers in the UK and Denmark. His research focuses on imaging techniques for nanoscience, industrial, and medical applications. A board member of the Spanish Microscopy Society, Lionel joined the University of Cádiz in 2016, contributing to the Intelligent Systems for Computation and Applied Magnetism and Optics groups.

Abstract:

Visual monitoring in Neonatal Intensive Care Units (NICUs) is a crucial clinical practice that allows medical staff to assess the health and development of preterm infants and optimize neonatal care. This observation includes evaluating periods of pain or stress, lethargy, abnormal movements, and adjusting environmental variables such as noise, light, temperature, and humidity, as well as monitoring vital signs. However, reliably identifying infants' emotions or specific patterns of motor activity remains challenging, episodic, and subjective, requiring the expertise of skilled clinicians. It is also time-consuming and susceptible to operator fatigue, skill variability, and inter-rater reliability issues. Vital signs are observed sporadically and not consistently recorded, and environmental comfort is rarely evaluated.

Due to these limitations, non-contact smart monitoring technologies based on video, audio, and other sensors are being extensively researched and are likely to become ubiquitous in future NICUs. In this talk various sensing technologies are described, with a focus on video monitoring using Edge AI (Artificial Intelligence at the edge) for NICUs. Edge AI involves running AI algorithms and processing data locally on hardware devices (edge devices) rather than relying on centralized cloud-based systems. This approach offers several significant advantages for medical applications, including reduced latency, enhanced privacy and security, and increased reliability. These benefits help meet the stringent technical and ethical regulations required in NICUs.



Turin, 08 - 10 July 2024





Neurodevelopmental outcomes prediction based on EEG abnormalities

Fabio Magarelli^{1,2,3}

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- 2. The SFI Centre for Research Training in Artificial Intelligence, CRT in AI.
- 3. Department of Pediatrics and Child Health, University College Cork, Ireland. fabio.magarelli@ucc.ie

Short bio of speaker(s):

Fabio Magarelli is a third-year PhD student at the Centre for Research Training in Artificial Intelligence (CRT-AI), University College Cork (UCC). He is supervised by Prof. Geraldine B. Boylan at the Infant Research Centre in Cork and Dr John O'Toole. His research focuses on predicting seizures in newborns using EEG data. He developed a machine learning competition platform and hosted a competition to classify background EEG abnormalities in newborns with Hypoxic-Ischemic Encephalopathy (HIE). Currently, he is investigating the temporal evolution of HIE within the first 100 hours from birth and its potential to predict neurodevelopmental outcomes at two years.

Abstract:

A comprehensive review of recent literature on EEG for predicting neurodevelopmental outcomes at two years, with a significant focus on babies with Hypoxic-Ischemic Encephalopathy (HIE). A brief discussion of a recent investigation of the temporal evolution of EEG background abnormalities in babies with HIE and how this could be used to predict outcomes by leveraging long-duration EEG recordings from previous multi-centre studies.



Turin, 08 - 10 July 2024





Advanced Bioinformatics for Medical Challenges

Seferina Mavroudi¹

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Short bio of speaker(s):

S. Mavroudi is a lecturer with the Department of Nursing at the University of Patras Greece. She teaches at the Postgraduate Program "Informatics for Life Sciences (ILS)" and served as an adjunct lecturer at the Department of Computer Engineering and Informatics at the University of Patras. During her PhD studies she visited the Bioinformatics Center of the University of Pennsylvania as a visiting researcher. She has over 90 publications in international scientific journals, proceedings of international conferences and book chapters and is the co-inventor of two submitted patents. She is a member of the IEEE-EMBS Greece Chapter Board.

Abstract:

From discovering new biomarkers, and advancing drug discovery processes to personalizing treatment plans, bioinformatics is transforming modern medicine. It bridges the gap between complex biological data and actionable medical insights paving the way for a future where medicine is more personalized, predictive, and precise.

In this talk, we will give an introduction to omics technologies which have become an integral part of many biomedical and clinical research projects. We'll investigate the basic omics data analysis workflows including the different steps required to analyze bioinformatics datasets, in a healthcare and biological context, from preprocessing data to completing the analysis task, interpreting the results and visualizing them. We will discuss analysis steps for particular types of omics data, including Next Generation Sequencing and Proteomics data, but also mention why and when to go beyond single omics and perform multi-omics analysis.

Finally, we will discuss the synergy between AI and bioinformatics and explore the applications and the potential of prominent state-of-the-art Large language models (LLMs) in performing bioinformatics tasks. We will have a look at the future of the field and close with the "big question" whether AI will entirely take over the bioinformatics field.



Turin, 08 - 10 July 2024





Neuroimaging in Premature Infants: Current Medical Practice and Challenges

Giovanni Morana¹

1. Neuroradiology Unit, 'The City of Health and Science' Hospital of Turin, Italy. giovanni.morana@unito.it

Short bio of speaker:

Giovanni Morana is Director of the Neuroradiology Unit at 'The City of Health and Science' Hospital of Turin and Associate Professor of Neuroradiology at the University of Turin, Italy. Since 2016 he is Fellow of the International Cancer Imaging Society (ICIS).

Past Chair of the European Society for Pediatric Oncology (SIOPE), Imaging Brain Tumor Group.

Currently he is Chair of the Pediatric Neuroradiology Section of the Italian Association of Neuroradiology (AINR).

He has published more than 130 papers in peer-reviewed medical journals indexed on Pubmed and 15 book chapters.

Abstract:

Developments in perinatal treatment over the last two decades have contributed to a decrease in the severe neurological morbidity and mortality rate of extremely and very preterm neonates. However, this population frequently has motor and/or cognitive problems linked to mild-tomoderate white and gray matter brain damage.

Injury from hypoxia, hemorrhage, or inflammation is particularly dangerous for the developing preterm brain.

Central to the assessment of premature infants is identifying the presence and extent of both mild and more severe forms of brain injury as well as many of the maturational disturbances of cerebral white matter and gray matter structures, using non-invasive neuroimaging techniques. Among neuroimaging techniques, the use of magnetic resonance imaging (MRI) has shed more light on injury patterns and particular vulnerabilities.

The main limitation of MRI relates to its availability for the neonatal population and the accurate interpretation of findings by experienced pediatric neuroradiologists.

The evaluation and depiction of the most frequent neuropathological patterns and MRI findings in preterm infants with perinatal brain injury is the focus of the present work.



Turin, 08 - 10 July 2024





Explainable AI and Uncertainty Quantification in Medical Applications

Silvia Seoni¹

1. Biolab, PoliTo^{BIO}Med Lab, Department of Electronics and Telecommunications, Politecnico di Torino, Corso Duca degli Abruzzi 24, 10129 Turin, Italy - <u>silvia.seoni@polito.it</u>

Short bio of speaker(s):

Dr. Silvia Seoni is a Research Assistant at the Department of Electronics and Telecommunications, Politecnico di Torino, Italy. She obtained her Ph.D. from Politecnico di Torino and University of Turin. Currently, she is actively involved in two European Horizon 2020 Research projects: Inside and REAP. Dr. Seoni has authored multiple papers in scientific journals and contributed to conference proceedings, including the IEEE International Ultrasound Symposium.

Her research primarily focuses on signal analysis, biomedical image reconstruction, and the application of Explainable AI (XAI) techniques and uncertainty quantification (UQ).

Abstract:

Artificial intelligence (AI) models have achieved excellent performance, comparable to human decision-making, especially in the healthcare sector. Due to their nature, these models are often seen as "black boxes", which generate a prediction from a given input without providing any explanation of how this prediction is decided and the uncertainty of the prediction itself. Explainable AI (XAI) is a set of techniques, models, and algorithms aimed at providing information on how the model generates its prediction, improving understanding and interpretability. However, these techniques do not allow for the evaluation of the model's reliability or robustness and the predictions. Therefore, uncertainty quantification is essential to assess the reliability of the model (in case of epistemic uncertainty) or the data (in case of aleatory uncertainty).

This talk provides a comprehensive overview of both XAI and UQ methods, focusing on their synergistic application in clinical settings, particularly in biosignal and bioimage analysis. Practical examples will illustrate how these techniques can improve image reconstruction, enhance pathology classification, and analyze biomedical signals, ultimately facilitating more reliable AI applications in real-world clinical scenarios.



Turin, 08 - 10 July 2024





CNNs for medical imaging and fetal neurodevelopmental diagnostics

Lemana Spahić¹

1. Lemana Spahić, Research Institute Verlab for Biomedical Engineering, Medical Devices and Artificial Intelligence, Sarajevo, Bosnia and Herzegovina. <u>lemana@verlabinstitute.com</u>

Short bio of speaker(s):

Dr. Lemana Spahić is a Doctor of Philosophy in Bioengineering and a senior expert associate at Research Institute Verlab, as well as a Marie Curie PhD Fellow at the University of Kragujevac. Her research interests include biomedical engineering and artificial intelligence with a profound emphasis on applied artificial intelligence in medical devices. With a firm background in biology and medicine she is a truly interdisciplinary scientist involved in development and implementation of a variety of projects ranging from medical device performance assessment to disease diagnosis decision support systems in healthcare. Up until now she has published more than 60 scientific contributions.

Abstract:

Fetal neurological impairment disorders, encompassing conditions like cerebral palsy, epilepsy, and autism spectrum disorder, can result from various factors affecting fetal nervous system development. Timely diagnosis of these disorders is challenging but crucial for early intervention. Recent advancements in deep learning and ultrasound technology present an opportunity to develop a tool for early detection. The study utilized a dataset of 3D ultrasound images extracted from 4D recordings of fetuses undergoing the Kurjak Antenatal Neurodevelopmental Test during the third trimester and convolutional neural networks (CNNs) to analyze fetal neurobehavioral movements in ultrasound images, with the goal of aiding in the early detection of neurological impairment disorders. The custom CNN architecture achieved an overall accuracy of 93.83%. The system was visualized by means of designing a graphical user interface that includes the developed model that works in the background every time a frame of a recorded 4D ultrasound video is deemed to be parsed through the system. Notably, distinguishing between facial and hand-to-face movements proved challenging. This pilot study lays the foundation for AI-based fetal neurological risk assessment, providing a promising tool for the early detection of fetal neurological impairment disorders. While acknowledging limitations such as class imbalance, the study demonstrates the potential of AI in enhancing prenatal care. Future work will involve expanding the dataset, conducting realtime clinical validations, and further refining the model. The research holds implications for improving outcomes for affected children and making advanced diagnostic capabilities accessible in diverse healthcare settings.



Turin, 08 - 10 July 2024





Artificial Intelligence based Decision Support Systems in Medicine

Michela Sperti¹

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Short bio of speaker:

Michela Sperti is a Ph.D. candidate at Politecnico di Torino, Bioengineering Department. She graduated in Biomedical Engineering at Politecnico di Torino in 2019 with a thesis on machine learning techniques for cardiovascular risk prediction in rheumatic patients. She worked for one year as a Research Assistant under the European-funded MSCA VIRTUOUS project (which aims to apply machine learning techniques to investigate taste and food properties). She is currently studying explainability techniques for machine learning models applied in clinical decision support systems with a special focus on cardiovascular risk prediction.

Abstract:

Risk assessment is fundamental for developing current medical treatment protocols, aiding in predicting future disease mortality and morbidity at both population and subgroup levels. *Personalized risk estimates*, which must also be self-explainable, are foundational to personalized medicine (a concept that emerged in the late 1990s). The concept refers to the ability to provide custom care to patients based on differences in response to a given disease or treatment. This approach requires novel techniques to handle and interpret the large and complex datasets it is based on.

Simultaneously, computer science and especially artificial intelligence progress allows now to store and process intricate and large datasets efficiently (a feat previously unattainable by early computing technologies). Within this context, machine learning (ML) aims to uncover complex patterns in data. These patterns can then be used for advanced exploratory data analysis, prediction, or classification of new and unseen data. However, a significant challenge in ML is the interpretability of models, as both input data and models become increasingly complex.

Current ML research focuses on enhancing model interpretability and reducing their *black-box* nature. This is central to clinical decision-making, as it enables clinicians to comprehend the rationale behind ML-driven decisions that impact patient care. Furthermore, interpretability will be essential for clinical trust and acceptance of ML models in medicine.

This talk aims to show how to build personalized, explainable, reliable, dynamic, and usable clinical decision support systems, progressing from traditional statistical methods to transparent ML approaches.



Turin, 08 - 10 July 2024





Introduction to -omics and related data analysis

Konstantinos Theofilatos^{1,2}

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Short bio of speaker:

Dr Konstantinos Theofilatos, is a Computer Engineer with an MSc and a PhD in bioinformatics from the Computer of Engineering and Informatics of the University of Patras. Since 2013, he co-founded and is the CTO of InSyBio, a multi-awarded biotechnology company focusing on the analysis of biomedical data using machine learning and network analytics techniques. Since 2017, he has been a Lecturer of Bioinformatics at King's College London. The research of Dr Theofilatos focuses on machine learning, bioinformatics, proteomics and multi-omics data analysis with applications in cardiovascular research and other domains. In particular, he has more than 10 years of experience in bioinformatics, biological network and machine learning data analysis (Düzgün MB, et al. OMCL. 2019; Theofilatos K, et al. BMCMG. 2019; Corthésy J, et al. JPR. 2018; Korfiati A, et al. EMBnet J, 2017; Theofilatos K, et al. AIM, 2015; Kleftogiannis D, et al. IEEE/ACM TCBB. 2015; Rapakoulia T, et al. Bioinformatics, 2014.) while for the last seven years, he has emphasized applying these techniques for the identification of therapeutic, prognostic and predictive biomarkers for cardiovascular diseases (Theofilatos et al, Circulation Research 2023, Moreira LM, et al. Nature, 2020; Schulte C, et al. Cir Res. 2019.). Dr Theofilatos has co-authored more than 45 publications with 2209 citations so far. He has co-edited 2 books and he is the primary inventor of two bioinformatics processes related to patent applications to the US patent office.

Abstract:

Omics techniques generate large, multidimensional data that require complex analysis by new informatics approaches alongside conventional statistical methods. Systems theories, including network analysis and machine learning, are suitable for analysing these data but must be applied with an understanding of the relevant biological and computational theories. In this hands-on workshop, we will cover all the available steps for performing analysis of Omics data including mass spectrometry proteomics and RNA-sequencing data. During the First part, we will focus on the preprocessing of raw mass spectrometry and RNA-sequencing using various normalization, imputation, filtering and scaling techniques. The first part will also include differential expression analysis and pathway/functional enrichment analysis. During the second part of the workshop, we will be working on the meta-analysis of the preprocessed data to perform optimized data visualization, clustering, train and test classification models and reconstruct and analyze molecular networks. The workshop will be implemented using the Google Colab platform (https://colab.google/) to execute coding Notebooks in R and Python with the attendees being called to customize the given codes in a series of questions. In the end, there will be devoted timing for the attendees to try these techniques for their own data.



Turin, 08 - 10 July 2024





AI for neonatal amplitude-integrated aEEG-based predictions

Tamara Škorić¹

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Short bio of speaker(s):

Tamara Škorić works as an Associate professor at Department of Power, Electronic and Communications, Faculty of Technical Science, University of Novi Sad, Serbia. Her scientific interests are focused on application of information theory and artificial intelligence in biomedical signal processing.

She is the Principal Investigator of one national project and the Vice-Chair of a COST action Al4NICU. She was the TCP and Chair for IoTH session at the IEEE ICC conference (May 2023). She was elected as a regional delegate (Croatia, Cyprus, Slovenia, Serbia, Bosnia and Herzegovina, Albania, Greece, Israel, and North Macedonia) and a member of board of EURACON.

Abstract:

During the lecture, a machine-learning model for the prediction of later seizure development in infants with hypoxic-ischemic encephalopathy, based on an Adaptive Boosting classifier will be explained in detail. The proposed model was supported by only two features extracted from amplitude-integrated electroencephalography aEEG (after the first 12 hours of recording) and six clinical data available from the newborn's history charts. All features were extracted in the time domain, and don't require the performing of standard raw electroencephalography (EEG). The limitations imposed by the model developed based on small bases (larger datasets are not available) as well as used equipment from different manufacturers will be discussed from the point of view of a fair comparison of available methods in modern literature. Data acquisition with equipment that does not represent the gold standard of medicine (for example Olympic Medical CFM Viewer 6000 device), but is being used in middle-income and low-income countries, is a real challenge and will be discussed.



Turin, 08 - 10 July 2024





AI Models for Brain Images and Eye-Tracking in Premature Children

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Short bio of speaker(s):

Andrea De Gobbis is a PhD student at the University of Ljubljana working at NEUS Diagnostics. He is researching the application of AI models to diagnostic procedures based on eye-tracking, focusing on adaptive testing methods to identify cognitive delay in premature children. Jaime Simarro is a PhD student at KU Leuven and a research engineer at icometix. His field of experience is related to Artificial Intelligence (AI) techniques applied to neuroimaging. Emiliano Trimarco, PhD at the UCA and INIBICA, studies neurodevelopment disorders. Roa'a Khaled, PhD student at the University of Cádiz (UCA), develops models for cerebral ultrasound segmentation. Marta Malavolta, PhD student at the University of Ljubljana (UL), develops ML algorithms with measurement uncertainty. Monica Crotti, PhD student at KU Leuven, studies brain damage in children with cerebral palsy. Arnaud Gucciardi, PhD at the UL and Toelt AI lab, explores computer vision for clinical images.

Abstract:

Preterm birth can cause long-term neurodevelopmental disorders due to prenatal or perinatal brain damage, such as cerebral palsy (CP) and autism spectrum disorder (ASD). Besides motor problems, children with CP present visual impairments for which early detection is crucial. Clinical neuroimaging techniques attempted to establish a relation between specific brain damage and visual impairment, and suggest the need for more advanced methods. Al-based software can significantly enhance the interpretation of neuroimages and eye-tracking data. Developing a fully automated quantification model for early childhood brain structures using MRI and/or cranial ultrasound could provide objective measurements of brain development, leading to better early detection and prognosis of neurodevelopmental disorders. For example, symmetry-aware neural network pipelines inspired by existing manual tools can be trained and evaluated on preterm MRI. Moreover, advanced voxel-by-voxel MRI analysis, in the form of Brain Age Gap Estimation, has shown potential in differentiating toddlers with ASD from their typically developing peers by capturing subtle structural variations that a standard volumetric approach cannot. Lastly, eye-tracking technology provides innovative tools for assessing cognitive impairment in preterm infants. We have created a neuropsychological test battery with engaging elements to maintain children's attention, improving data quality and engagement in clinical trials. In conclusion, analyzing neuroimages and eye-tracking data in preterm infants requires tools and models that go beyond current methods. Integrating AI with pediatric brain signals could help fill the current gap and enhance early diagnosis strategies.



Turin, 08 - 10 July 2024





Bioinformatics in biomarker discovery in preterm infants

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Short bio of speaker(s):

Arantxa Ortega is a PhD student at the University of Cádiz and an early-stage researcher at the PARENT project. Her research focuses on the application of machine learning techniques for predicting neurodevelopmental outcomes in very preterm infants using clinical, image-based, and omics data. **Konstantinos Panagiotopoulos** is a PhD student at Politecnico di Torino, Italy and a research fellow in the European project PARENT. His field of experience is Bioinformatics and particularly transcriptomics combined with Machine Learning. He is currently focused on developing models for the identification of biomarkers and the homogenization of datasets to enhance the analyses. **Lolia Ibanibo** is a PhD student at the University of Cadiz and researcher at the research institute INIBICA, and one the early-stage researchers of the MSCA PARENT project. She has a background in Molecular Biology and prior experience as a Biomedical Scientist. Lolia is working to discover potential circulatory microRNA signatures to be used as non-invasive biomarkers of all forms of white matter injury, a comorbidity which is prevalent in the premature infant.

Abstract:

White matter injury (WMI) is one of the most prevalent damages in preterm infants (PTIs), often leading to long-term neurodevelopmental impairments (NDI). Although neuroimaging techniques can detect macrocystic WMI, diffuse WMI, a more subtle and prevalent form, is not easily detected. In some cases, diffuse WMI remains undetected until the child starts showing signs of NDI. Recent studies indicate that microRNAs (miRNAs) participate in oligodendrocyte development, myelin formation, and the pathogenesis of WMI. MiRNAs as biomarkers are excellent diagnostic assays. We propose that the expression pattern of circulating miRNAs in the peripheral blood of premature newborns is related to the development of white matter lesions and subsequent NDI. Our working group employs a bioinformatics approach, using publicly available transcriptomics datasets to build a well-tailored repository to enhance analyses. This helps us narrow down potential miRNA and other biosignatures, indicative of healthy or NDI in PTI. Using prior knowledge, we validated 20 miRNAs in 30 PTIs using qPCR. Three miRNAs showed differential expression between PTIs with WMI and no WMI. Furthermore, preliminary results from NGS data analysis obtained from six patients, have identified differentially expressed miRNAs in PTIs with WMI and no WMI. Specifically, 64 miRNAs were found to be upregulated and 228 downregulated. These differentially expressed miRNAs hold potential as biomarkers for early detection and could lead to the development of accessible diagnostic assays.



Turin, 08 - 10 July 2024





Advancing Pediatric Healthcare: Improving Outcomes in Congenital and Neonatal Care

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Short bio of speaker(s):

Enrico Piccinelli, MD, PhD candidate, is a clinical and interventional paediatric cardiologist at Ospedale Bambino Gesù in Rome. **Janet Pigueiras**, a Telecommunication Engineer and PhD student at Cadiz University, is working on motion and physiological patterns of infants. **Syed Adil Hussain Shah**, a Computer Scientist, is a PhD candidate at GPI SpA and Politecnico di Torino, working on vocal analysis in neonates. **Syed Taimoor Hussain Shah**, a Computer Scientist and PhD student at Politecnico di Torino, is researching time-based disease progression. **Pablo Romero**, a Telecommunication Engineer, is a PhD candidate at Politecnico di Torino and 7HC, designing precision medicine tools.

Abstract:

Innovative approaches in imaging, data collection, machine learning, and artificial intelligence (AI) are providing new insights and tools for healthcare professionals. Given the importance of early diagnosis, we are employing 4D imaging, real-time neonatal data, vocal analysis, facial recognition, data collection, and tools usage application platforms to assist surgeons. Pulmonary regurgitation (PR) after Tetralogy of Fallot (ToF) surgical repair is common. 4D flow MRI and computational fluid dynamics may help in predicting the pulmonary valve replacement timing with possible impact on clinical outcomes. In pediatric care, 4D flow MRI aids health monitoring, while AI tools support early medical decisions for infants. Increasing premature births necessitate specialized NICU monitoring. Continuous electronic medical records enhance diagnostics and outcomes. The Neonates Recording Platform (NRP) integrates physiological data, video, body pose analysis, and audio for comprehensive care. Voice emotion and vocal analysis tools use spectrogram and Xception Net transfer learning, validated on diverse datasets for robustness. Early neurological disease detection employs FRAI, a facial-feature-trained CNN model, aiding early-diagnosis and medical decisions, potentially reducing healthcare burdens. Both tools employ explainable AI techniques to elucidate relationships between source and target data. In addition to developed tools, PARENT DSS is a robust platform aiding researchers and doctors with predictions for early decisions. It supports model training from the PARENT Project, handles data augmentation, and manages health data, including diverse predictions such as child mood. These innovations in neonatal care, congenital heart disease and health data integration promise improved patient outcomes and informed healthcare delivery.