Milan Macek Jr.

Prof. Milan Macek Jr. MD, DSc, M.H.A.

Professor Milan Macek Jr. MD, DSc is the chairman of the largest academic medical / molecular genetics institution in the Czech Republic – Department of Biology and Medical Genetics of Charles University Prague-2nd School of Medicine and Motol University Hospital, and of the National Coordination Centre for Rare Diseases (www.nkcvo.cz; NKCVO) responsible for implementation of the ten year national strategy on rare diseases and resulting three national action plans. In addition, he is chairing the national Rare Disease Taskforce at the Ministry of Health. In this capacity his institute has been serving as a "clearing centre" for the dissemination of knowledge gathered within various international projects on rare disease-related research and diagnostics (e.g. EuroGentest.org, RD-Connect.eu, Solve-RD.eu, Norway Grants) to partners in Eastern Europe, Transcaucasia and the Middle East. In this capacity Prof. Macek is also the Czech National coordinator of Orpha.net. In his capacity as chairman of NKCVO he assured that since 2017 Czechia is ranking first within EU13 in terms of participation in European Reference Networks (ERN) for rare diseases.

Prof. Macek is also the past President of the European Society of Human Genetics (www.eshg,org; 2010-2011 ESHG), currently serves at the ESHG liaison for European National Human Genetics Societies (https://www.eshg.org/76.0.html). Under his leadership medical genetics was recognized as an official EU specialty in the Professional Qualifications Directive in 2011. He also closely collaborated with the Council of Europe on the ratification of the Additional protocol on genetic testing for health purposes to the Oviedo convention (2019). He had also been the past-board member of the European Cystic Fibrosis Society (ECFS.eu; 2007-2014) and is the current member of the European Cystic Fibrosis Registry board (https://www.ecfs.eu/ecfspr), whereby he published seminal papers on the disparities in cystic fibrosis care between the “New” and “old” EU Member States. Moreover, he had also been the board member of the European Society of Human Reproduction and Embryology (www.eshre.eu; ESHRE) where he was responsible for three joint position statements of ESHG and ESHRE in the field of reproductive genetics as their senior author. Prof. Macek served at the European Commission Expert Group on Rare Diseases (formerly www.eucerd.eu) and is currently involved in the European Board of Member States for European Reference Networks for Rare Diseases (https://ec.europa.eu/health/ern_en), including the newly formed EU Advisory Board on ERN sustainability. He had also been member of the Diagnostic Committee of the International Rare Disease Consortium (www.irdirc.org). Prof. Macek is currently the president of the Czech Society of Medical Genetics and Genomics (www.slg.cz). Finally, he was the chief government advisor of the CZ EU Council presidency under which the EU Council recommendation on a field of action in rare diseases was been adopted in 2009.

Prof. Macek did his postdoctoral studies at the Department of Medical and Human Genetics at Humboldt University Berlin (1989-1992) followed by McKusick-Nathans Centre for Genetic Medicine, Johns Hopkins University in Baltimore (1992-1996). In 1992 he was also a fellow at Harvard School of Medicine in the field of non-invasive prenatal diagnosis of rare diseases.

His main research and clinical interests are in the field of molecular genomics of rare diseases, including their deep phenotyping, and in ways on how to bring genomics knowledge to the bedside via targeted therapies with orphan medicinal products. He has also been involved in health economics “cost of illness” studies in this regard. His citation index is over 11,000x with H –index of 33.

Margarida D. Amaral

Margarida D. Amaral is Full Professor of Biochemistry/ Molecular Biology at the Faculty of Sciences, University of Lisboa (Portugal) and Coordinator of BioISI - Biosystems & Integrative Sciences Institute. MDA is alumna of EMBL-European Molecular Biology Laboratory (2008-10; 2016) and of IGC - Gulbenkian Institute of Science (1983-1993). EMBO member (2014).

Research: The Amaral lab has its major focus on the molecular and cellular mechanisms of biogenesis, traffic and degradation of normal and mutant protein CFTR, which when mutated causes the genetic disease Cystic Fibrosis (CF). To understand CF mechanisms globally we use transcriptomics, proteomics and functional genomics (functional siRNA screens). Our results translate into the clinic for better CF diagnosis, prognosis and personalized therapies. Author of 156 international peer-reviewed publications 4,662 citations, H-factor 38 (Scopus).

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https://ciencias.ulisboa.pt/en/perfil/msamaral

Isabelle Sermet-Gaudelus

Isabelle Sermet-Gaudelus is a professor in Paediatric at the Necker-Enfants Malades Hospital, Paris France. She is now the head of the paediatric Cystic Fibrosis center, at the Necker-Enfants Malades Hospital. She has been engaged for several years in a programme of phenotype-genotype studies, using epithelial electrophysiological measurements. She runs a clinic which accepts referrals from throughout France for questionable diagnosis for CF. She is involved in the European Clinical Trial Network as head of the standardization committee. She is the head of a research laboratory at INSERM whose main focus is CFTR interactome and molecular mechanisms for specific CFTR mutations.