**Hassen Hadj Kacem**, Eng, PhD,

Centre de Biotechnologie de Sfax, Tunisie

hassen.hadjkacem@cbs.rnrt.tn

Université de Sharjah, Faculté des Sciences,

Département de la Biologie Appliquée.

hkacem@sharjah.ac.ae

Hassen Hadj Kacem, est titulaire d'un doctorat en génie biologique de l'université de Sfax depuis 2004 et directeur de recherche au centre de Biotechnologie de Sfax depuis 2012. Il est actuellement Professeur Associé à l'université de Sharjah, coordinateur de l'équipe de recherche « Bio-informatiques et génomique Fonctionnelle » et membre de l'équipe de recherche « Génétique Humaine et Cellules Souches ». Dr. Hassen s'intéresse à l'identification des biomarqueurs des pathologies humaines héréditaires moyennant les technologies de génotypage et de séquençage de l'ADN à faible, moyen et à haut débit principalement la nouvelle génération de séquençage (NGS) et de l'étude de leurs effets fonctionnels. Durant les vingt dernières années Dr. Hassen s'est intéressé aux pathologies endocriniennes rares et a collaboré étroitement avec le service d'endocrinologie du CHU Hédi Chaker de Sfax dans l'analyse génétique des insuffisances antéhypophysaires isolées et syndromiques et les formes rares du diabète monogénique. Dans la suite, une liste courte des publications les plus significatives avec le service d'endocrinologie du CHU Hédi Chaker de Sfax durant les dix dernières années.

## JEANNE AMIEL

Jeanne AMIEL (PU-PH) is a clinical geneticist and leads since 2019 the team "Embryology and Genetics of Malformations" in INSERM U1163 affiliated to the *Imagine* Institute (https://www.institutimagine.org/fr/jeanne-amiel-75) . She has conducted a number of studies aiming at gene identification for neural crest cell derived congenital malformations and tumour predisposition, in particular in Hirschsprung disease, congenital central hypoventilation, neuroblastoma, conotruncal heart defects and mandibulofacial dysostoses. She is or has been the coordinator or associate investigator in a number of research programs funded by the French National Agency for Research (ANR) and the Fondation pour la Recherche Médicale (FRM). She is coordinating the *CRMR Anomalies du Développement* at Necker Hospital affiliated to the *Filière AnDDI-Rares* and the *Master Européen de Génétique* at Université de Paris.

## JOSE LUIS DE LA POMPA

José Luis de la Pompa is a developmental biologist who leads the « Intercellular Signalling in Cardiovascular Development & Disease Laboratory » at the Centro Nacional de Investigaciones Cardiovasculares Carlos III (CNIC) in Madrid since 2009. Most of his group's efforts have been centered on the role of NOTCH and interacting signalling pathways in cardiac valves and chamber development and disease. His group is currently studying how NOTCH signalling in the valves interacts with other pathways or genes to cause valve dysmorphology and has identified a genetic signature predictive of aortic valve calcification in human. In the cardiac chambers, his group studies how different pathways including NOTCH, regulate the cellular and molecular mechanisms involved in early ventricular development. They have also showed that NOTCH signalling disruption impairs the latter process of chamber compaction, causing left ventricular non-compaction cardiomyopathy (LVNC) in mice and human. Exome analysis of Left Ventricule non compaction (LVNC) families has identified a set of genes potentially interacting with NOTCH to cause LVNC. Work in progress aims at characterizing the phenotype of the corresponding mutant mice and the biochemical interactions among gene products involved. Mutations in some sarcomere genes cause both LVNC and Hypertrophic Cardiac Myopathy. The genetic and mechanistic bases of these apparently disparate cardiomyopathies are currently being investigated. He is a member of the ESC WG on Development, Anatomy & Pathology.

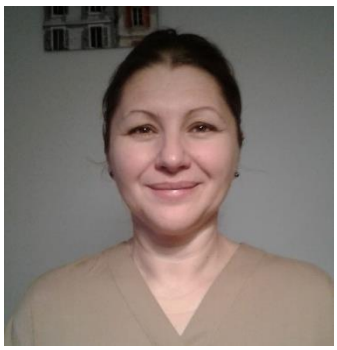
## ALBERTO M. PEREIRA

Alberto M. Pereira is professor of Medicine, and head of the Division of Endocrinology at the Leiden University Medical Center, in the Netherlands.

In 2012 he has founded and since then chairs the Center for Endocrine Tumors Leiden, an (inter)national multidisciplinary reference center for patient with endocrine tumors based on patient-centered care pathways and Value Based Health Care (VBHC) linking care and research. He was president of the European Neuroendocrine Association (2016-2018), and at present is the coordinator of the European Reference Network on Rare Endocrine Conditions (Endo-ERN). His research focusses on the long-term effects of pituitary tumors and of its treatment, and in specific on the effects of stress hormones on the central nervous system. He is the PI of the recently awarded European Rare dIsease research Coordination and support Action (ERICA) H2020 Project (29 partners): joining forces to integrate research and innovation capacity across all 24 European Reference Networks.

He authored more than 300 papers and book chapters (current h-index: 53), and has supervised 16 PhD candidates.

## MARIA POPA-ROCH



Maria Popa-Roch is currently an assistant professor in social psychology at the University of Strasbourg.

Her research activities are generally organized around the theme of social perceptions of individuals because of their group membership. More specifically, she is interested in the psychosocial mechanisms involved in the stigmatization of minority social groups, as well as its consequences.

Models of social cognition and those derived from social identity theory provide the theoretical basis for her research. In her recent work, she has been interested in the negative attitudes that children with disabilities face in educational settings, as well as the implications of these attitudes in the current context of inclusive education. The role of implicit and explicit stereotypes and prejudices is addressed in this work.

## XENIA PROTON DE LA CHAPELLE

Either for rare diseases or more widespread diseases, patient's organizations are increasingly present in the health ecosystem, in France and abroad, in order to support patients and their families in the long course of the disease, but also to publicize the diseases, develop communication between families, clinicians, researchers, and support research.

As the mother of a young child suffering from a rare disease presenting a vital risk, the Ondine Syndrome (also called CCHS, Central Congenital Hypoventilation Syndrome), Xenia entered 6 years ago into a world which was completely foreign to her, the world of diseases and disability. She thought back then that "it only happens to others," until the day it happened to her and her family as well, when her 4th child was born. A difficult journey then began, from understanding the disease to its daily management at home, including discussions with the doctors in charge of his medical care.

Since fatality is not part of her vocabulary, she has decided to get involved in improving the daily life of her son and the patients suffering from this very serious disorder. For a few years, she was the president of the Ondine family association (AFS Ondine) to help families sometimes in very precarious situations to manage their child at home, fight with the difficulties of the French administrations in order to benefit from financial support, have access to the presence of a dedicated nurse at school or simply reassure them about the daily life and future. Because when you are parents of a sick child, it is important to be able to imagine the future.

At the same time, the numerous discussions with researchers working on the Ondine syndrome gradually led her to understand how important and how hopeful research was, and after having provided funding to Ondine research teams via our french association, she finally created a start-up company last year whose mission is to develop a therapeutic solution for Ondine patients.