

Collaborative European centres participating to the EBRA cluster:

Centre for Genomic Regulation (CRG), Barcelona, Spain

Dr. Mara Dierssen (MD, PhD)

Dr. Dierssen's work is devoted to understanding the pathobiological bases of complex brain disorders such as Down syndrome with the aim of finding new cures that help the cognitive alterations and social inclusion of intellectually disabled patients. Her work in these last years has focused on boosting activity-dependent neural plasticity in the brain, using combined pharmacological and non-pharmacological treatments both in mouse models and in human clinical trials. She combines molecular biology, neuromorphological and behavioural studies in mice and humans, and mathematical modelling to decipher the relevant molecular pathways underlying treatment efficiency. She is founder of the Trisomy 21 Research Society and past president of the Spanish Society for Neuroscience and IBANGS. She has strong links with patients' associations.

Institut du Cerveau et de la Moelle (ICM), Salpêtrière Hospital, Paris

Drs. MC Potier (PhD) and JM Delabar (PhD)

Dr. Marie-Claude Potier has been working on DS for the past 20 years. She recently developed new treatments for cognitive deficits in DS, particularly by modulating GABAergic transmission using selective GABA α 5 inverse agonists. Dr. Jean Delabar is a worldwide expert on DS and the chromosome 21 gene DYRK1A. Both are founding members of the Trisomy 21 Research Society. Dr. MC Potier is the General Secretary and Dr. J Delabar is the Chair of the Sponsoring Committee. This group is conducting research on Alzheimer's disease (AD) in DS.

Radboud University Medical Centre (UMC), Radboud, The Netherlands

Dr. Tonnie Coppus (MD, PhD) As a medical doctor specialised in intellectual disability care and an epidemiologist, Tonnie Coppus started a longitudinal research project in 1999 on predictors of dementia and mortality in DS. In 2007, she developed a specialised national multidisciplinary outpatient department for adults with Down syndrome. Tonnie Coppus was one of the founders of the Dutch multidisciplinary Down Team Research consortium (DOC), a joint venture of representatives of the Dutch Down Syndrome Patient Organisation and health care and research professionals. Since 2012, she has been heading the Radboud University Nijmegen research pillar "Down syndrome".

Consorti Mar Parc de Salut (IMIM), Barcelona, Spain

Dr. Rafael de la Torre (PharmD, PhD).

Co-ordinator of Human Pharmacology and Clinical Neurosciences Research Group and Director of the Neuroscience Research Programme at IMIM. Full Professor of Toxicology at Universitat Pompeu Fabra (CEXS-UPF). He has 284 indexed publications (h index 48), and he has supervised 22 PhD students. He has coordinated clinical trials in Down syndrome and other intellectual disabilities. Dr. Vicky Puig is also part of this lab. She is investigating the neural substrates of cognitive impairment in mouse models of DS.

Centre Européen de Recherche en Biologie et Médecine (CERBM), Illkirch, France

Dr. Yann Herault (PhD) Dr. Herault aims to understand the pathophysiology of DS investigating mouse models in order to go further and develop therapeutic approaches to circumvent the consequences on cognition. As such we developed and continue to produce DS mouse models for large homologous genetic region and for genes of interest.

We focus our interest on cognition in young adult mice but we also explore other alterations observed in DS people. We contributed to the validation of a few drugs that are now explored in clinical trials with the aim to recover some cognitive performance in DS people. We also started to investigate late phenotype in DS models.

University College London (UCL), London, UK

Prof. Lizzy Fisher (PhD)

Our group has worked on DS for over 20 years by creating novel animal models to help dissect pathological pathways. We are currently focused on three main areas, the development of dementia, heart defects, and abnormalities of the central nervous system, and the models we create have been used in pre-clinical trials.

University of Antwerp with Alzheimer Research Centre (ARC) Groningen, Antwerp, Belgium

Prof. Peter Paul De Deyn (MD, PhD)

Scientific director of the Institute Born-Bunge, Antwerp Belgium and the Alzheimer Research Centre in Groningen, The Netherlands. De Deyn has ample experience with national and international collaborations related to both preclinical and clinical research in the Alzheimer field (recently also very much involved in Alzheimer's Disease in DS). The close collaboration between the research laboratories and the clinical departments contributes to the thorough know-how and expertise of the complete research centre and facilitates comparison of the human condition and relevant animal models. An important focus is on pathophysiological substrates of Behavioural and Psychological symptoms of Alzheimer's disease and Down Syndrome – Alzheimer's Disease. Efforts have been devoted to unravelling pathophysiological mechanisms of neurodegenerative disorders such as dementia and to research on CSF biomarkers in MCI and dementia. Substantial efforts have been put into validation of a range of AD mouse models and possible therapeutic interventions, involving the application of acetyl cholinesterase inhibitors, neuroprotectants, secretase modulators and beta-amyloid vaccination as well as gene therapy. As member of this cluster we look forward to achieve synergisms between DS and Intellectual Disability experts and AD experts. Dr. Alain Dekker has recently graduated from Prof. Peter De Deyn's lab.

University of Bologna, Bologna, Italy

Prof. Renata Bartesaghi

Dr Bartesaghi's research group is focusing on the molecular mechanisms underlying intellectual disability in Down syndrome in view of therapeutic approaches during early life stages aimed at rescuing the brain developmental alterations that characterize Down syndrome. Dr. Sandra Guidi is a Researcher at the School of Medicine in Bologna. She worked for about ten years on detecting the mechanisms underlying brain hypotrophy in DS. Her current focus is to find a possible treatment potentially useful to improve neurogenesis impairment also in humans with Down syndrome. Dr. Fiorenza Stagni is a postdoc in the lab of Prof. Bartesaghi.

University of Cantabria, Santander, Spain

Prof. Carmen Martinez-Cue (PhD)

Dr Martinez Cue main interest is in DS neuropharmacology. Her main reasons for willing to join this network are to establish collaborative research on intellectual disabilities and cognitive impairments with other groups and to promote new research on intellectual disabilities for IMI.

University of Geneva, UNIGE, Switzerland

Prof. Stylianos Antonarakis (PhD)

Antonarakis laboratory is interested in the understanding of the molecular pathophysiology of the phenotypic heterogeneity of trisomy 21. We use cellular systems and genome analysis to study the differences in genome function in trisomy 21 and also the contribution of the genomic variability to the phenotypic variability. In addition, our clinical recruitment of patients with trisomy 21 and their families provide the possibility for translational, clinical research projects.

Kings College London, UK

Prof. Andre Strydom (MD, PhD)

Prof. Strydom is a very active clinical researcher and practitioner in the field of DS. His research concerns the epidemiology, aetiology and clinical aspects of mental disorders in individuals with neurodevelopmental disorders including intellectual disability (ID) syndromes such as Down syndrome (DS), fragile X syndrome (FXS), and Autism Spectrum Disorders, with a major focus on Alzheimer's disease in Down syndrome. Prof. Andre Strydom has extensively demonstrated his scientific leadership in the field is Chair of the Dementia in Intellectual Disabilities special interest group, he leads the LonDownS research collaboration, a multidisciplinary research programme on the neurobiology of Alzheimer's pathology in DS, he is Advisor to the London Research Design Service and Member of the UCL Rare Diseases Steering Committee.

Hospital de Sant Pau, Barcelona, Spain

Dr. Juan Fortea (MD, PhD)

Dr. Fortea has been developing biomarkers in DS and in DS-AD. Ha has also constituted cohorts of DS patients with a biobank of samples including plasma, CSF and cells. His main goal is to understand the development of Alzheimer's disease in DS to predict those individuals who will convert to AD for, in the future, propose new therapies able to delay and/or combat AD.

University La Sapienza, Roma, Italy

Dr. Marzia Perluigi (PhD)

Assistant Professor at the Department of Biochemical Sciences "A. Rossi-Fanelli", Sapienza University of Rome. She is actively involved in DS and Alzheimer Disease research area. She is the supervisor of the laboratory of redox biochemistry in neuroscience at Sapienza University, Department of Biochemical Sciences and has developed new tools to study organelles in the context of DS.

University of Cambridge, Cambridge, UK

Prof. Pietro Lio (PhD)

Professor of Computational Biology at the Department of Computer Science and Technology of the University of Cambridge. He has great expertise in deep learning and artificial intelligence and has developed new algorithm to compute big data in many diseases including Alzheimer's disease to predict clinical features and disease evolution. He is very motivated to apply these new techniques in the field of DS.