

Thematic Workgroup on Down syndrome research priorities: Research infrastructures and biocollections



On November 29th, 2021, the TRISOMY21-cluster came together in Barcelona (Spain) with representatives from EU research infrastructures, networks, cohorts, repositories, biobanks, patient associations, and relevant stakeholders, from funding agencies to industry. They discussed European infrastructures for DS research and biocollections.

In the morning, several EU Cohorts and Registries presented their work including Horizon 21 Consortium, the independent cohorts/bio sampling initiatives la Princesa Madrid, PoliClinico Gemelli Rome, French DS cohort Lyon, Defeating Dementia in Down Syndrome (DIDS) Cambridge, Down Alzheimer Barcelona Neuroimaging Initiative (DABNI), Genetic Forms of Intellectual Disability and Autism Spectrum Disorders (GENIDA), and the European network of population-based registries for the epidemiological surveillance of congenital anomalies (EUROCAT). This was followed by presentation from the EU Biobanks: BioJEL Jerome Lejeune Foundation, LonDowns, European Bank for induced pluripotent stem cells (EBiSC), Telethon and Askion. Finally, they discussed together 1. How to structure registries/repositories/biocollections for DS research; 2. How to incorporate all existing cohorts/samples in large efforts such as Horizon 21; 3. Issues with regards to collection and storage of relevant samples for DS research and 4. The need for harmonization and guidelines for biobanking in DS research.

In the afternoon, similar initiatives in the US (i.e., Alzheimer's biomarker consortium for Down syndrome (ABC-DS), The Alzheimer's Clinical Trial Consortium-Down Syndrome (ACTC-DS), the INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome (INCLUDE) Project and the Data Management and Portal for INCLUDE (DAPI) project shared their knowledge and experience on how to set up, contribute to and use registries/repositories/biobanks. Then, the European Strategy Forum on Research Infrastructures (ESFRI) and Biobanking and BioMolecular Resources Research Infrastructure-European Research Infrastructure Consortium (BBMRI-ERIC) presented services and support they can offer to the Down Syndrome research community. The day finished with a round table open to all participants.

The meeting provided a clear view on the European landscape of research infrastructures in Europe. A lot of work has already been done but there is still too much fragmentation and the accompanying challenges. A roadmap has been created on how to move forward but we can capitalize on what has already been done for Down Syndrome research in the US. We need to start building Down Syndrome Research Infrastructures across the borders and coordinate the work in Europe and globally. The TRISOMY21 cluster will play a very important umbrella role in this effort.