Genetic diseases

We study the pathogenetic mechanisms of Kabuki Syndrome and related pathologies, called Chromatinopathies, using cellular models combined with biochemical and bioinformatic models. Our primary goals are: i) to profile the DNA methylation signature in a larger cohort of Chromatinopathies patients, in order to use it as molecular diagnosis tool, ii) to study the functional interplay between alteration of topology associated domains (TADs), and transcriptional profiles in Kabuki syndrome patients, and iii) to identify the role of KMT2D-mediated epigenetic modifications related to metabolism. By combining biochemical and metabolomic approaches in mouse and human cell models, we demonstrated a rewiring of the mitochondrial metabolic phenotype due to KMT2D alteration that might contribute to the onset of Kabuki syndrome. We currently using our cellular models to go deeply in the biological mechanisms underpinning this metabolic control.
Finally, we have Biobanked about 800 individuals with Kabuki syndrome and related chromatinopathies, of which we have a careful clinical description, Primary cell lines such as skin fibroblasts and stem cell lines such as hiPSC have been generated and made available for the international scientific community.

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