

LES LAURÉATS 2021

LES LAURÉATS DE L'APPEL À PROJETS 'GENOMICS'

RESPONSABLE DU PROJET	VILLE	APPROCHE TECHNIQUE	TITRE DU PROJET
Minchul Kim	Strasbourg	10X Genomics single nuclei RNAseq	Tracking the origin of tubular aggregate myopathy by single-nucleus transcriptomics
Djihad Hadjadj	Paris	Hi-C chromosome conformation capture	Identification of constitutional genetic abnormalities predisposing to Familial midgut carcinoid tumors
Marion Delous	Lyon	RNA seq	Transcriptomic Study During Neuronal Differentiation Of Rnu4atac-Associated Rare Diseases: Connecting Splicing Defects To Brain Developmental Abnormalities
Stéphane Viville	Strasbourg	WES	Deciphering genetic causes of non-obstructive azoospermia via exome sequencing; a way to personalize treatments and to develop new diagnostic tools.
Audrey Putoux	Lyon	WES	Towards the identification of genes involved in recurrent anencephaly
Marie-Louise Frémond	Paris	WES	Identifying Novel Monogenic Interferon-Mediated Juvenile Rheumatoid Arthritis
Xavier Puéchal	Paris	WES	Deciphering genetic and molecular bases of Whipple disease
Sandrine Vuillaumier-Barrot	Paris	WGS	Research of homozygous variant in 5' or 3'UTR ALG12 region or cis regulating elements by whole genome for one ALG12-CDG suspected patient
Celine Huber	Paris	RNAseq	Identification of new pathogenic variants in individuals with Chondrodysplasias with multiple dislocations (CMD) combining WGS and RNASEQ