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GOUVERNEMENT
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LE GRAND PLAN D'INVESTISSEMENT

Programme prioritaire de recherche
Maladies rares

Résultat de l'appel à projets 2020 Maladies rares : résoudre les impasses diagnostiques

Coordinateur du projet	Acronyme et titre du projet
CHARRON Philippe	**ResDiCard** Resolving diagnostic deadlock in Cardiomyopathies
DOLLFUS Hélène	**CIL-RET PASS** Solving variants of uncertain significance (VUS) in patients with ciliary-related retinal dystrophies by designing comparative phenotypic and transcriptomic tools generated from iPSC & retinal organoids
DUPONT Jean-Michel	**CHROMAPS** Prospective study to assess medical performance of optical mapping and long read sequencing in detecting numerical and structural chromosome abnormalities
POGGI Marjorie	**BioFIT** Biomarkers of inherited thrombocytopenia
RENDU John	**CLASSIF AI RYR1** Integrated Classification of RYR1 Variant of Unknown Significance
REYNIER Pascal	**PRIOMICS** Deep omics phenotyping for genomic variants prioritization
SOUMELIS Vassili	**CD-STROMA** Deciphering the lymphoid stromal cell network in Castleman disease, for development of new diagnostic tools and innovative therapeutic targets
TOURNIER-LASSERVE Elisabeth	**GENETWORKDIAG4** Development of novel strategies based on Gene Network. Computational Approaches to identify causal genes in rare Cerebral Small Vessel Diseases
VITOBELLO Antonio	**MultiOmixCARE** Neurodevelopmental disorders and negative short-read genome sequencing: multi-omics approaches to trace a roadmap towards the diagnostic management of post-genome investigations to reduce wandering and diagnostic deadlock

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maladies rares