

Appel d'offres “ Plateforme Mutations ”

Résultats Réunion du Conseil Scientifique 21 Janvier 2010

Liste des projets sélectionnés (ordre alphabétique)

AMIÉL Jeanne	Identification of the disease causing gene in Late-Onset Central Hypoventilation Syndrome with hypothalamic dysfunction (LO-CHS-HD)
COLLEAUX Laurence	Identification of a gene(s) responsible for Cornelia de Lange syndrome
LE BER Isabelle	Whole exome sequencing in autosomal dominant forms of frontotemporal dementias
LE GOFF Carine	Identification of a gene(s) responsible for Acromicric Dysplasia
LYONNET Stanislas	Identification of the disease causing gene in Kabuki syndrome
MARLIN Sandrine	A new gene for dominant isolated deafness

THAUVIN-ROBINET Christel

Search for identification of a gene responsible for a new syndromic X-linked mental retardation with severe microcephaly and dystonia

SOUBRIER Florent

Identification of novel gene(s) involved in Hereditary Haemorrhagic Telangiectasia

VIDAUD Dominique

Exomic sequencing for identification of the gene responsible for a neurofibromatosis 1 phenotype