FONDATION maladies rares

High Throughput Sequencing and Rare Diseases
Call for Proposals
'GenOmics of rare diseases'

Assumptions *(tick if appropriate)*:

Monogenic disorder ☐ Genetic heterogeneity: Yes ☐ Likely ☐ No ☐

Digenic disorder ☐ Oligogenic disorder ☐ Multifactorial disorder ☐

Mitochondrial transmission ☐

Mode of inheritance of the disease

Ascertained ☐ Anticipated ☐ Unknown ☐

Autosomal dominant ☐ Autosomal recessive ☐ X-linked dominant ☐ X-linked recessive ☐

De novo mutation(s) Yes ☐ No ☐

Complete penetrance Yes ☐ No ☐ Incomplete penetrance Yes ☐ No ☐

Variable expressivity Yes ☐ No ☐ Phenocopies Yes ☐ No ☐

Other:

Preliminary data *(tick if appropriate)*:

- Have large genomic rearrangements been excluded (CGH arrays…)? Yes ☐ No ☐
- Have all known disease related-genes been tested? Yes ☐ No ☐
- Previous exome sequencing on the same disease? Yes ☐ No ☐

Comments:

- Previous exome sequencing on the same samples? Yes ☐ No ☐

Comments:

Cases history *(tick if appropriate)*:
For human exome or genome sequencing, please indicate:

- Familial Yes ☐ No ☐
  - Consanguineous ☐ Non consanguineous ☐
  - Large ☐ Small ☐ families Number of families:
  - Number of patients in each family (nbx/Fx; nby/Fy; etc):
  - Available DNA for: - patients (nb): - parents (Y/N):
  - Genome wide linkage analyses performed: Yes ☐ No ☐ Not feasible ☐

- ✓ Conclusive linkage Yes ☐ No ☐
- ✓ If yes, specify: - the region delineated (and size):
  - the maximum lod score obtained:

- o Sporadic Yes ☐ No ☐
  - Consanguineous ☐ Non consanguineous ☐ families
  - From a clinically homogeneous cohort Yes ☐ No ☐
  - Number of patients:
  - Available DNA for: - patients (number): - parents (Y/N):

Technical approach	**Number of samples**
Whole exome	
Whole genome	
RNA-seq	
small RNA-seq	
ChIP-seq	
Methyl-seq	
Single cells	
Other, please specify:	

Total number of requested samples for the present project: ☐