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300 EXOMES TO ELUCIDATE RARE DISEASES

Submission Form

1. Project

1.1 Title

1.2 Acronym

1.3 Primary phenotype Choose an option

If other, please specify

2. Principal Investigator

2.1 Name and surname

2.2 ID Card

2.3 Position Choose an option

If other, please specify

3. Institution

3.1 Name

3.2 Department, Group or Service
(please specify name of the Head)

3.4 Post Code, City

4. Five most relevant publications, preferentially as last author, by the applicant in Genetics / Genomics/Clinics of Rare Disorders

5.Description of the project

5.1 Introduction - Disease context, disease prevalence, current knowledge about causative mutations/variants, available diagnostic tests, etc. (300 words maximum).

5.2 Mode of inheritance

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

Genetic Heterogeneity

- ☐ Yes
- ☐ Likely
- ☐ No

Complete penetrance

- ☐ Yes
- ☐ No

Phenocopies

- ☐ Yes
- ☐ No

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5.3 Cases history

5.3.1 Familial or Sporadic (choose option A or B)

☐ A) Familial

Consanguinity

☐ Yes ☐ No

Pedigrees

☐ Nuclear ☐ Extended

Number of families ☐

Number of available* PATIENT DNA samples per family (average) ☐

Number of OTHER available* DNA samples per family (average) ☐

☐ B) Sporadic

Consanguinity

☐ Yes ☐ No

From a clinically homogenous cohort

☐ Yes ☐ No

Number of available* DNA samples from PATIENTS ☐

Number of available* DNA samples from PARENTS ☐

*** >6 ug good quality DNA samples MUST be available at the time of submission**

5.3.2 Brief description of the Patients/Families available for the study (150 words maximum).

* If working with extended families, please enclose a separate pdf file with the pedigree structures indicating available DNA samples and appropriate legends.

5.3.3 DNA samples

Source

☐ Blood ☐ Saliva ☐ Hair ☐ Biopsy ☐ FFPE ☐ Other

If other, please specify

Quality

☐ Good quality ☐ Some smear ☐ Bad quality/degraded

Amount available (measured using absorbance methods - nanodrop or equivalent)

☐ 6-10 ug ☐ >10 ug

5.3.4 Relevant clinical information (100 words maximum).

Clinical/Phenotype description available using Human Phenotype Ontology (HPO) terms

☐ Yes ☐ No

5.3.5 Relevant karyotype / array CGH / linkage / expression / candidate gene sequencing results for these samples, if available (100 words maximum).

5.3.6 Previous testing of all known disease related-genes

☐ Yes, all ☐ Yes, most of them ☐ Yes, some of them ☐ No

5.4 Proposed analysis plan (candidate genes, linkage peak regions, mutation type, etc.) (150 words maximum)

5.5 Potential follow-up experiments (sequencing additional patient samples, animal models, in vitro studies, etc.) (150 words maximum)

The researcher _____

agrees to be the principal investigator of the submitted project, as it is described in the present application, and confirms that:

- The samples were obtained with the corresponding approval of the Bioethics Committee and signed "informed consent" from each donor, both for collection and for their use, including conservation, manipulation/sequencing by entities such as CNAG.

- The "informed consent" allows that anonymized samples and sequencing data and results are included in CNAG's internal databases and distributed in secure controlled access databases such as the European Genome-phenome Archive (EGA) .

- The applicant does not hold any award directly related to Whole Exome Sequencing in the same group of disorders.

☐ Mark this box to accept the terms and conditions

At _____ *on the* _____ *in the year 2013.*