



## Manifestation of interest

### Characterization of tissue type I interferon involvement in renal disease of monogenic type I interferonopathies

**Project short name:** KID\_interferon

#### **Brief rationale:**

Monogenic type I interferonopathies (T1I) (like COPA syndrome (COPA), STING-associated vasculopathy with onset in infancy (SAVI) or some monogenic lupus) may display renal involvement. These entities are characterized by a systemic upregulation of type I interferons (IFN-I) which is central to the disease pathogenesis. However, a clear documentation of IFN-I upregulation at tissue level is still lacking. Indeed, this would be crucial to understand the selective organ involvement encountered in these diseases. The demonstration of tissue IFN-I upregulation can be performed applying human myxovirus resistance protein 1 (MxA) immunostaining to renal biopsies. While this characterization has been assessed in a few patients with T1I-related kidney diseases, it has never been performed in SAVI, COPA and several others T1I.

#### **Objectives:**

- 1- To demonstrate by immunostaining the presence of MxA up-regulation in kidney biopsies of patients with COPA- and SAVI-related renal disease (or other monogenic T1I) in terms of percentage of marked area/total area.
- 2- To describe by immunostaining the tissue-localization of MxA and its co-localization within specific cell types and to explore the association between the staining pattern and the different histopathological kidney lesions encountered in COPA and SAVI patients (or other monogenic T1I).
- 3- To compare MxA percentage of marked area/total area, tissue-localization, co-localization within specific cell types of COPA and SAVI kidneys (or other monogenic T1I) with those of sporadic lupus nephritis (LN)
- 4- To identify the cellular sources of IFN-I in the kidney through in situ hybridization on kidney biopsies.

#### **What we need from you:**

- 1- Unstained slides from paraffin-embedded kidney biopsies and/or kidney biopsies in paraffin-embedded tissue blocks from patients with SAVI or COPA-related renal disease or other monogenic T1I or monogenic lupus. This material will have to be sent to the Nephrology Unit of the Meyer Hospital in Florence, Italy together with the pathology report and routine fluorescence images.
- 2- Fill out a Clinical Report Form with clinical and laboratory information of the patients.

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#### **Scientific Partners**

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