



# ITHACA Board Meeting

2020 December 10 – 12

Satellite Meetings : Friday Dec. 11<sup>th</sup>

WG 7 Research

Follow-up on research collaborative call: Pai/OAFNS

Solve  RD



European  
Reference  
Networks

# Collaborative call – Pai OAFNS



- First call as soon as the collaborative call initiative was launched in May

**Targeted gene(s)/phenotype under study:** Pai/OAFNS syndrome, ideally after negative exome

**Abstract :** The Solve-RD project is aiming at identifying genes in diseases resistant to exome sequencing. Pai and OAFNS are overlapping syndromes, belonging to the unsolvable cohorts in solve-RD, taking into account that exomes have remained negative in a number of patients. Multiomics are proposed to identify the molecular bases of these entities. New samples are required, including blood, urine and saliva of the proband and parents, and ideally tissue of affected area if available.

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**Specific requirements beyond clinical data and genotype data sharing:**

- Re-analysis of DNA samples : **N**
- Resampling of patients No if the trio is available : **Y**
- Linked to a translational/basic research project? **Y, Solve-RD**

## Pai syndrome and oculo-auriculo-fronto-nasal syndrome (OAFNS) hypotheses

### Constitutional monogenic disease

Coding or non-coding (regulatory) variation not detectable by short read sequencing

- *Long read sequencing*
- *RNA-seq*
- *Proteome*
- *Epigenome*

### Mosaic disease

Variation not detectable in blood

- *Tissue analysis*

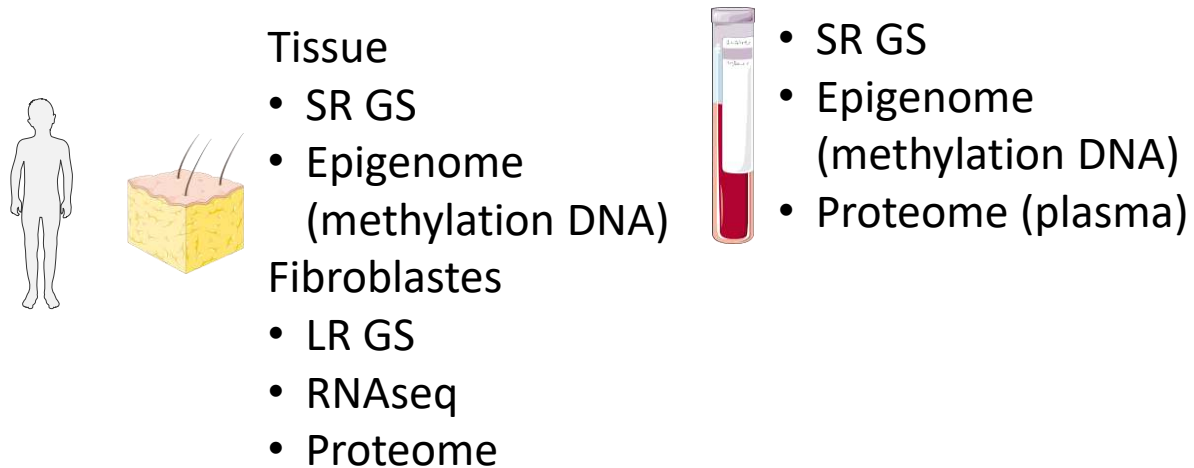
### Oligogenic disease

Multiple genes belonging to the same pathway

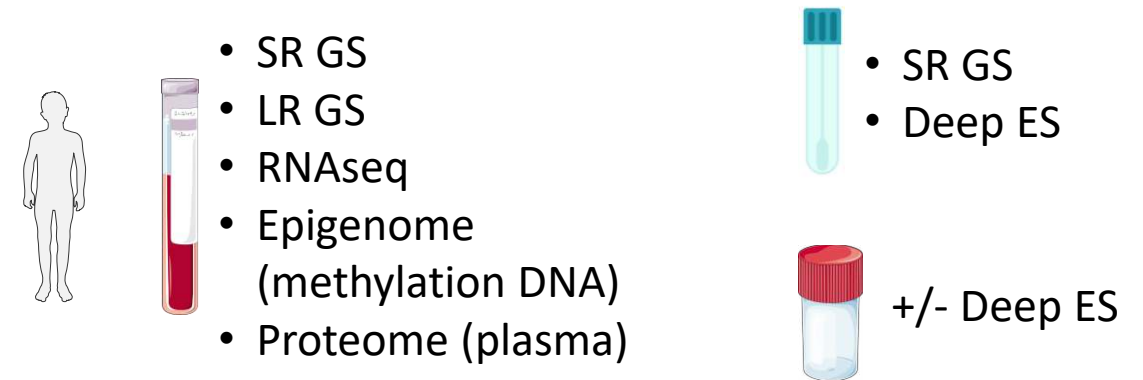
- *Multomics data integration*

## Analysis strategy for the Pai syndrome and OAFNS

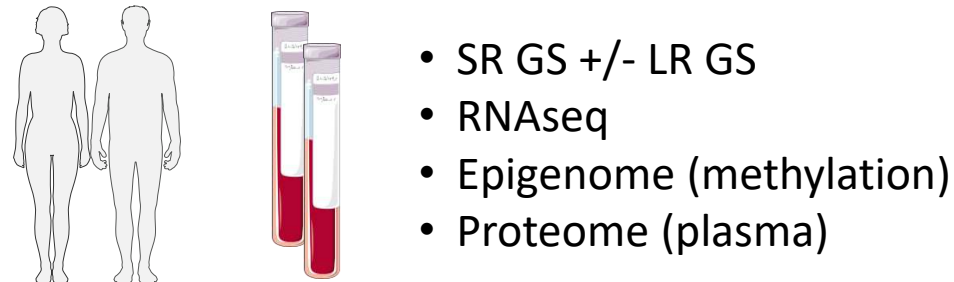
### Affected tissue available



### No affected tissue available



SR : short-read  
LG : long-read



# (WG8) Work Groups / Satellite Meetings : Friday December 11th



	France	ITHACA
Pai/OAFNS included with tissue	4	0
Pai/OAFNS included with no tissue	4	0
Pai/OAFNS with no samples sent	5	8 (UK 2, Italy 1, Spain 1, Netherlands 1, Cyprus 1, New Zealand 2)
Replication cohort	5	3