

“Study Of Secondary Cancers In Patients With Hereditary  
Retinoblastoma; Design Of A Strategy Based On Liquid Biopsy  
For An Early Diagnosis And Adequate Follow-Up  
(RET-LB Project)”

EURbG Meeting

London June 2023

Led by:

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Hereditary Cancer Research Group

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President of NDU Foundation

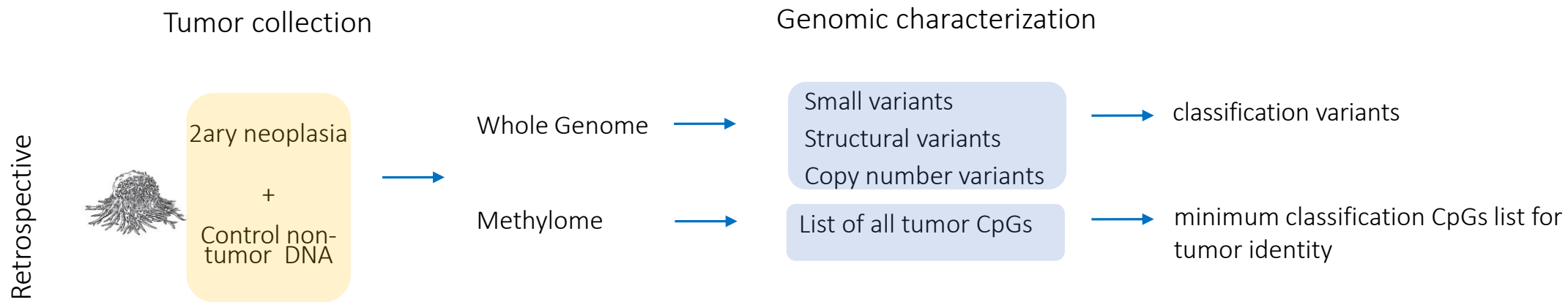


## The problem we would like to face: **secondary malignancies**

- Improvements in the treatment and follow-up of **hereditary retinoblastoma** children have resulted in a growing population of adult **survivors**.
- However, hereditary retinoblastoma survivors have a significantly increased risk of **secondary malignancies**. This is a major concern for patients and families.
- Their onset follows a somehow ordered temporal logic, conditioned by the type of therapy received previously for retinoblastomas.
- Some of these tumors are: leukemias, Ewing sarcoma, osteosarcomas, leiomyosarcomas and other soft-tissue sarcomas, melanoma, and epithelioid tumors.

# What we propose to do: **NGS-based test in liquid biopsy for early detection**

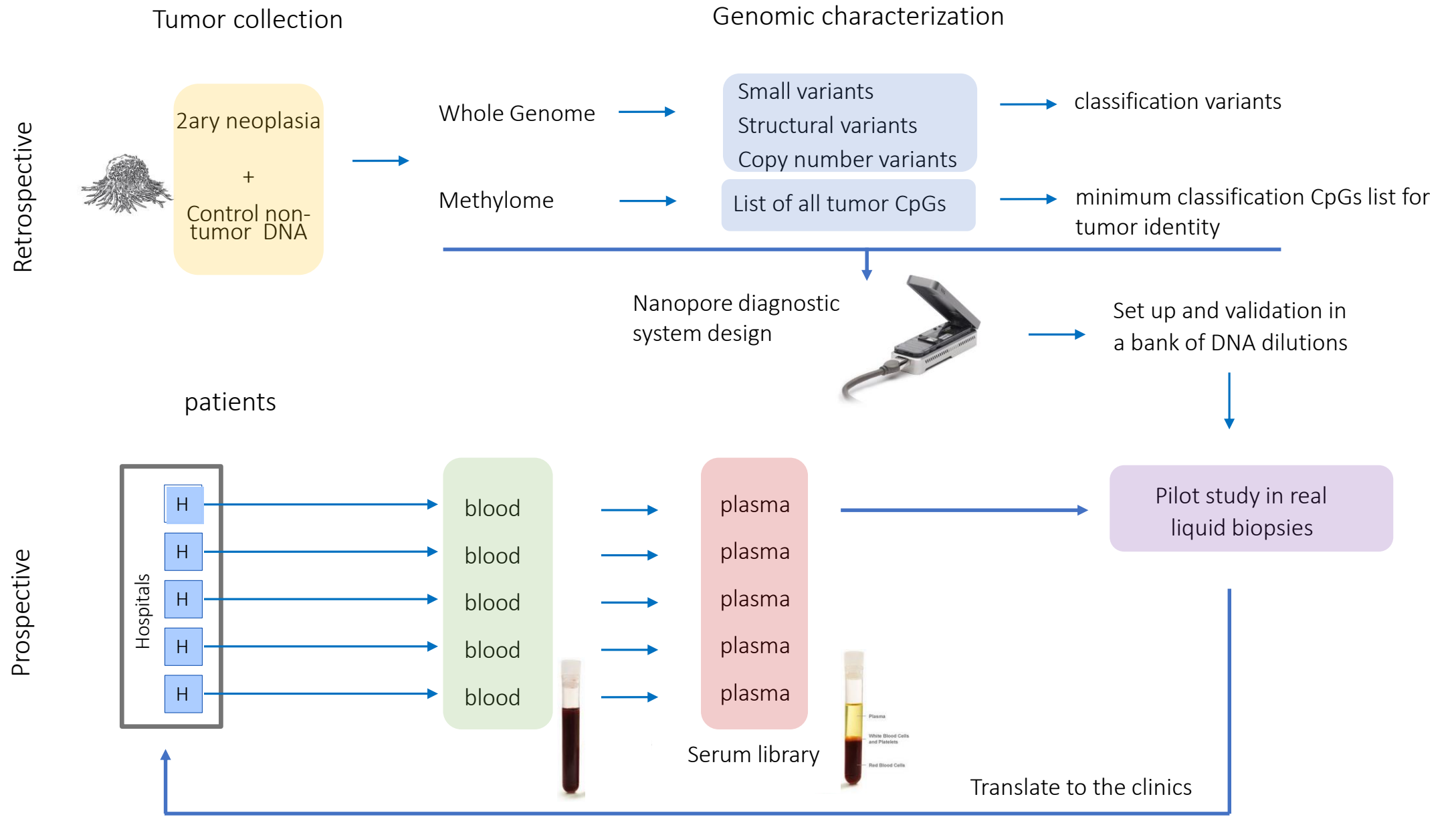
- Generate a genomic and epigenomic radiography of the most frequent secondary neoplasia appearing in the context of hereditary retinoblastoma.
- Develop a combined genomic and epigenetic test using nanopore NGS sequencing for a differential diagnosis.
- Perform a proof of concept analysis using DNA mixtures resembling real proportions of tumor DNA in plasma.
- Analyze real liquid biopsies from patients with hereditary retinoblastoma.



- Obtain a list of genomic regions and a list of CpGs to set up the test
- This information must allow the identification of all secondary neoplasia characterized
- This information must allow the differential diagnostic among them.

Nanopore  
diagnostic  
system design





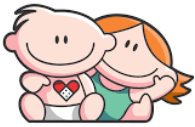
## Current teams involved

**SJD**

Sant Joan de Déu  
Barcelona · Hospital

**LaFe**  
Hospital  
Universitari  
i Politècnic

**ICO**  
Institut Català d'Oncologia



Hospital de Pediatria  
**Garrahan**

**Vall d'Hebron**

**FUNDACIÓN VALLE DEL LILI**  
*Excelencia en Salud al servicio de la comunidad*

**GRAACC**

Hospital  
**IOP**  
Instituto de  
Oncología Pediátrica

Parceria técnico-científica  
**UNIFESP**  
UNIVERSIDADE FEDERAL DE SÃO PAULO  
1911

**HUM**  
Hospital Universitario  
Virgen Macarena

**IGTP**  
Institut de Recerca Germans Trias i Pujol

## Open to collaborate/participate

### ● In the retrospective part:

Share genomic data on secondary neoplasia  
Provide samples, either DNA or tissue

### ● In the prospective part:

Share your experience in liquid biopsy  
Share your experience in liquid biopsy sequencing  
Provide liquid biopsy samples

### ● In any other way you like

### ● We can offer:

Open to incorporate new teams into the project  
Open to share genomic data produced  
Provide genomic analysis of new samples

Fundació  
**LA NINETA  
DELS ULLS**

## Contact

More information:

### Research issues

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### Organization issues

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