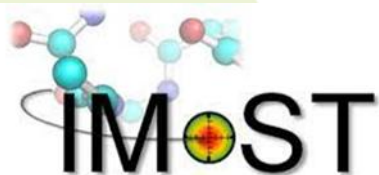


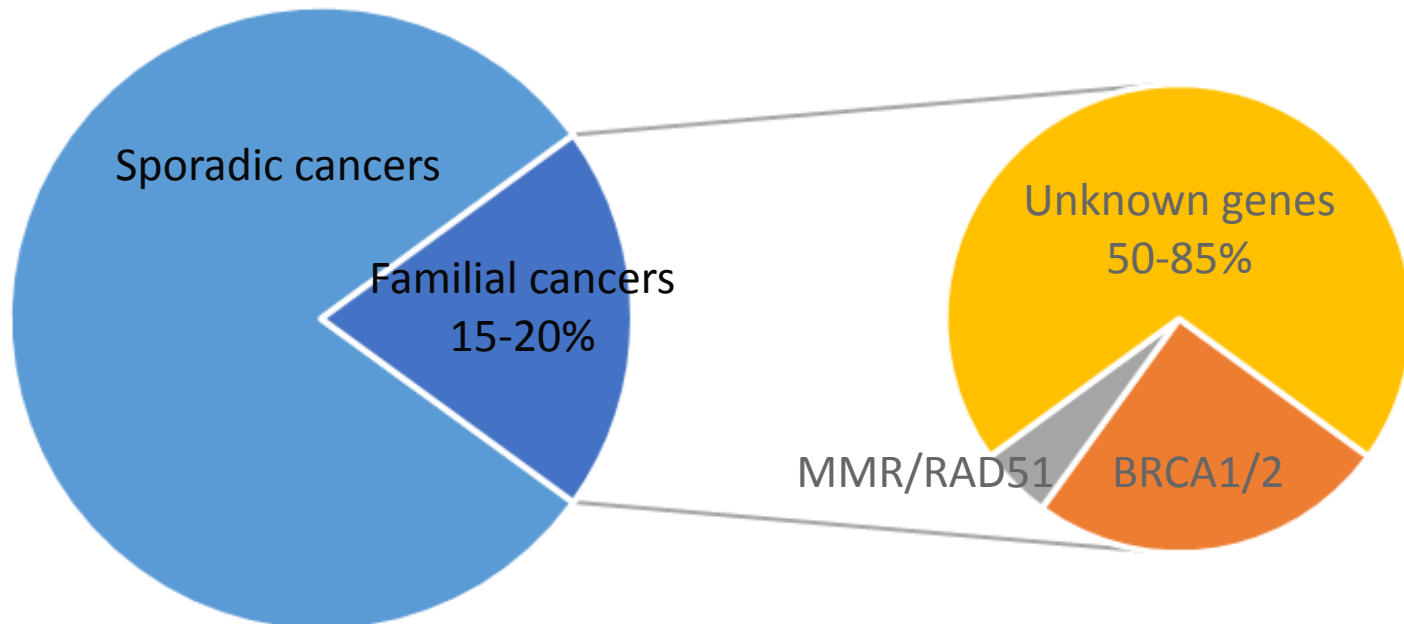
# Proposal of new candidate genes of predisposition to serous ovarian cancer using whole-exome-sequencing of 16 patients with a familial form

Mathias Cavallé, Maud Privat, Lorenzo Menicucci, Flora Ponelle, Nicolas Sonnier, Sandrine Viala, Nancy Uhrhammer, Yannick Bidet, Yves-Jean Bignon.



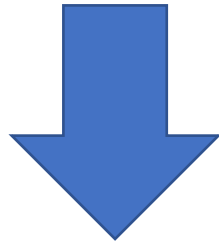
# High grade serous ovarian cancer

- ◆ 9th type of cancer
- ◆ 5th cause of death
- ◆ Late diagnostic and poor prognosis
- ◆ 15 - 20% of familial cancers :



# Constitutional exome sequencing

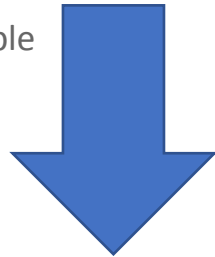
11 000 families in oncogenetic department of Centre Jean Perrin



- 2 cases of high grade serous ovarian cancers ;
- No breast cancer ;
- No mutation in BRCA/RAD51/MMR genes

16 families of hereditary predisposition of high grade serous ovarian

30 -50000 variants/sample



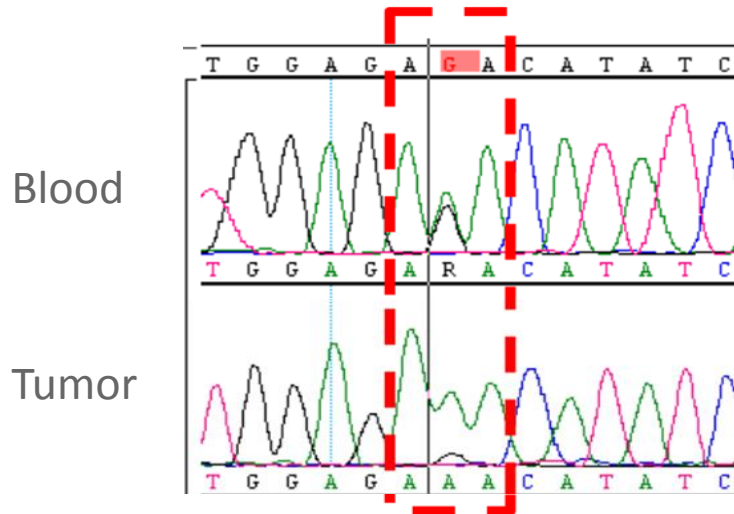
- Rare variants (GnomAD <0,5%) ;
- Truncating variants or splice variants or missense variants
- Genes with variants in at least 2 families ;
- Genes described in carcinogenesis processes

37 variants in 12 candidate genes implicated in 3 pathways

# Confirmation of the pathogenicity of missense variants

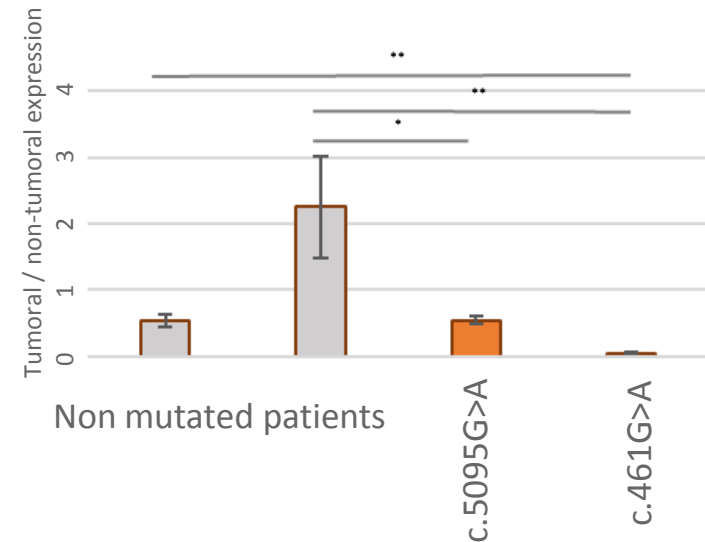
## Loss of heterozygosity

DLEC1 c.461G>A

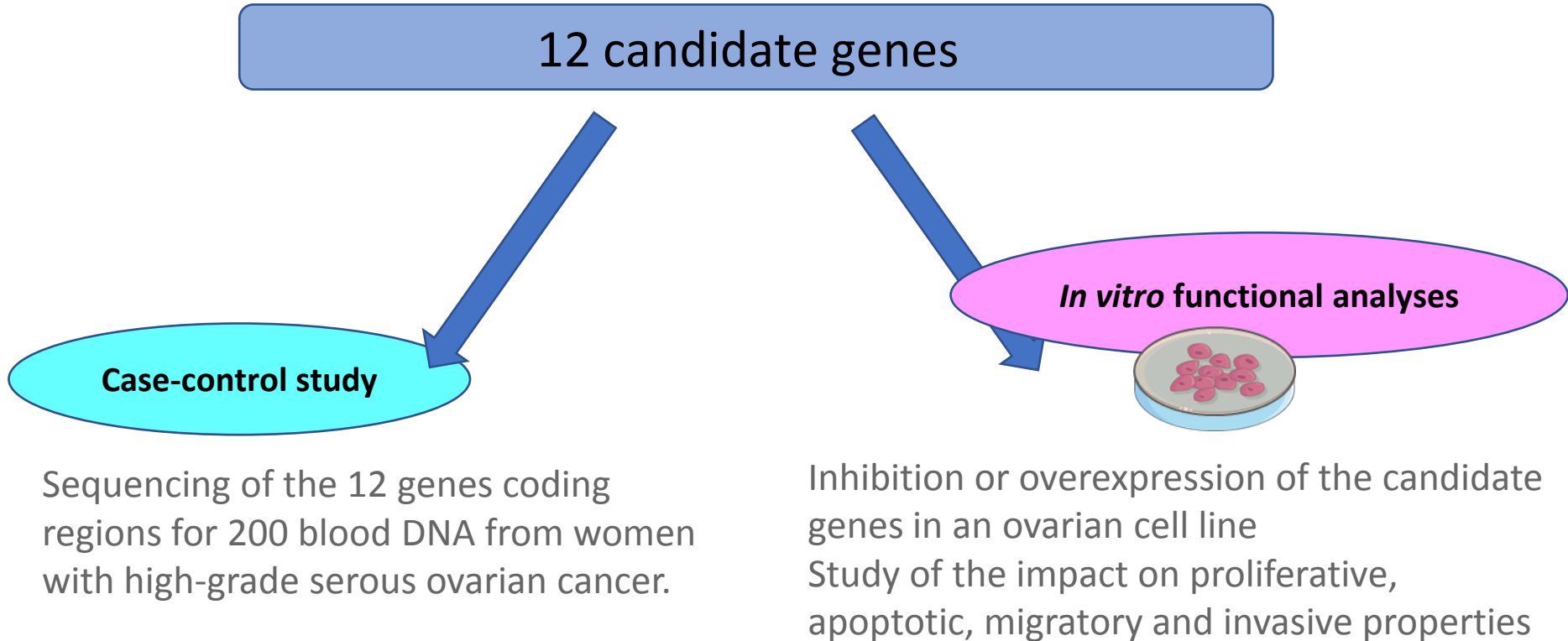


## Tumoral expression

DLEC1 tumor suppressor



# On-going experiments



**Objective : Identification of a new predisposing gene of high-grade serous ovarian cancer**