Our Group is mainly interested in identifying genetic risk factors involved in endocrine tumour susceptibility. Through a comprehensive analysis of tumour genomic features, we have been able to propose diagnostic and prognostic markers, to identify altered pathways that could be therapeutically targeted, and to identify new major susceptibility genes.

We are also interested in defining markers associated with differences in antitumour drug response and toxicity. We are applying targeted and whole-exome next-generation sequencing to a large series of clinically well-characterised patients. The aim is to identify new therapeutic approaches to personalise cancer treatment. These efforts will collectively improve the diagnosis, prognosis, and treatment of patients.

“We identified NOP10 as a new metastatic risk marker for paraganglioma, and recommend genetic screening in patients with metastatic non-clear cell renal cancer, as they show high prevalence of pathogenic germline mutations in renal cancer-predisposition genes.”
Analysis of telomere maintenance related genes reveals NOP10 as a new metastatic-risk marker in Pheochromocytoma/Paraganglioma (PPGL)

One of the main problems we face with PPGL is the lack of molecular markers capable of predicting the development of metastatic PPGL. To date, a limited number of markers have been proposed, such as TERT, SLC39A6, and H3.3, which have been shown to be associated with metastatic PPGL. However, the use of these markers has not been validated in a clinical setting. In this study, we aimed to identify a novel telomere-related gene that could be used as a potential metastatic marker for PPGL.

We performed a comprehensive analysis of telomere-related genes in PPGL using cDNA microarray analysis. We identified NOP10, a gene encoding a protein involved in telomere maintenance, as a novel metastatic marker for PPGL. NOP10 expression was found to be significantly higher in metastatic compared to non-metastatic PPGL samples. In addition, NOP10 expression was found to be associated with shorter time to progression and increased risk of disease recurrence.

Conclusion: Our findings suggest that NOP10 could be a new metastatic marker for PPGL and may have potential clinical applications in the management of PPGL patients.