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We use genomics-based approaches to study the genetics of cancer with focus on characterizing interactions between oncogenic pathways and the identification of novel cancer-related genes. Our research model is a hereditary neural crest-derived tumor known as pheochromocytoma or paraganglioma. These tumors are highly vascular, catecholamine-producing neoplasms that have been fundamental in providing insights into more common cancer forms and, because of their genetic heterogeneity, have served as sources for discovery of novel oncogenes and tumor suppressor genes. Using integrative genomic approaches, we recently identified a novel pheochromocytoma susceptibility gene, TMEM127, which functions as a classic tumor suppressor gene. This gene encodes for a transmembrane protein of unknown function.

Main research lines in the lab are related to:

- Characterizing the function of the novel tumor suppressor gene TMEM127, using in vivo (recently developed mouse model) and in vitro approaches with focus on TMEM127 involvement in mTOR signaling and endosomal function;
- Discovery of novel cancer genes using genomic and next-generation sequencing strategies, exploiting an available databank of well-annotated tumor samples;
- Identifying translational correlates of genetically defined tumor types that might guide the design of personalized care strategies for cancer patients.

Recent publications:

1. Yao et al. (2010) Spectrum and prevalence of FP/TMEM127 gene mutations in pheochromocytomas and paragangliomas, JAMA. 2010 Dec 15;304(23):2611-9.
2. Qin Y et al. (2010) Germline mutations in TMEM127 confer susceptibility to pheochromocytoma Nat Genet 42, 229-33
3. Yao L, et al. Mutations of the Metabolic Genes (2010) IDH1, IDH2,and SDHAF2 Are Not Major Determinants ofthe Pseudohypoxic Phenotype of Sporadic Pheochromocytomas and Paragangliomas J Clin Endocrinol Metab Mar;95(3):1469-72.
4. Schlisio S et al. (2008) The kinesin KIF1Bbeta acts downstream from EglN3 to induce apoptosis and is a potential 1p36 tumor suppressor. Genes Dev 22: 884-893.
5. Dahia PLM et al. (2005) A HIF1a regulatory loop links hypoxia and mitochondrial signals in pheochromocytomas. PLoS Genet 1: e8.