Genetics and Pathogenesis of congenital hypogonadotropic hypogonadism (CHH) and other disorders of puberty

The discovery of new genes involved in congenital hypogonadotropic hypogonadism (CHH) may allow for a better genotype-phenotype correlation, as well as an improvement in genetic counseling.

Mutations present in new genes identified by this project will increase our understanding of neuro-endocrine control of reproduction with potential applications to a broader spectrum of fertility pathologies. Through the doctoral thesis, the candidate will develop a variety of genetic applications focused on high-throughput sequencing data (e.g. WGS, RNA-seq., single cell transcriptomics) to discover new genes and/or pathways involved in the pathogenesis of congenital hypogonadotropic hypogonadism. As an MD, the candidate will also be involved with the phenotyping of CHH patients.