The **Molecular Pathology/ Complement Genetics** Group led by Prof. Santiago Rodríguez de Córdoba develops a multidisciplinary approach to decipher the molecular basis of rare human disease and to develop diagnostic and therapeutic strategies.

Their first works focused on **Alcaptonuria**, a rare autosomal recessive disease characterized by a disorder of the metabolism of tyrosine and phenylalanine. They solved their molecular, epidemiological and structural bases, marking the beginning of medical genetics and turning this work into a classic cited in genetics textbooks and popular scientific publications. Shortly after, they cloned the genes that cause 3-methylcrotonylglycinuria, another metabolism disorder, and SIX6, a gene involved in anophthalmia and pituitary abnormalities. They also contributed to identify one of the two genes that cause **Lafora disease**, a fatal form of progressive myoclonus epilepsy, and in subsequent work, using biochemical approaches, cell biology and animal models, defined the main pathogenetic mechanism of this disease.

In addition, the group has made important contributions to the genetics and biology of complement proteins, becoming an international leader in the study of this fundamental component of innate immunity. They have described the pathogenic mechanisms of two rare diseases associated with dysregulation of complement, **atypical Hemolytic Uremic Syndrome** (aHUS) and **C3 glomerulopathy**, justifying the implementation of therapies based on complement inhibitors.

All this research work has laid the foundations for the organization of a diagnostic laboratory for the study of complement in rare renal diseases (DCOM), which has become an international reference in the field of nephrology. DCOM provides a complete genetic and molecular diagnosis of the complement system in patients with complement-related disorders in support of a personalized medicine.