ALMA MATER STUDIORUM
UNIVERSITÀ DI BOLOGNA

DIPARTIMENTO
DI FARMACIA
E BIOTECNOLOGIE

AVVISO DI SEMINARIO

Il giorno **26/03/2026**
alle ore **12:00**

Prof. Xavier De La Cruz

Full Professor - Vall d'Hebron Research Institute - Barcelona (Spain)
(ospite del Prof. Emidio Capriotti)

terrà un seminario in lingua inglese dal titolo:

Understanding the Molecular Basis of Hereditary Disease in the Age of AI

in presenza:
Via Beverara 123 - UE1 - Aula 1F (Piano Primo)

Personale docente e ricercatore, dottoranti, assegnisti e studenti LM sono cordialmente invitati

ABSTRACT

Hereditary diseases are often caused by genetic variants that alter the function of key proteins. Understanding the functional impact of these variants is therefore a central challenge in human genetics and essential for explaining the molecular basis of many inherited disorders.
In this talk I will present our work on AI-based models designed to predict the functional consequences of genetic variation. By integrating evolutionary, structural, and biochemical information, these approaches help address a problem that is difficult to resolve through experimental or manual analysis alone and are becoming increasingly important for interpreting human genomic variation.
I will also briefly reflect on how generative AI tools are beginning to support the research process itself, acting as an always-available collaborator that helps explore complex questions and accelerate scientific reasoning.

BIOGRAPHICAL SKETCH

My research focuses on the development and application of computational approaches to address biological and biomedical questions. During my Ph.D. I studied the structural principles underlying protein function, work that I continued at the National Institutes of Health (1993–1997) and at University College London (1997–2000). After joining ICREA, this line of research remained central to my activities at the Parc Científic de Barcelona (2001–2009) and later at IBMB-CSIC (2009–2012).
Over time my interests have shifted toward translational problems in biomedicine. In 2012 I joined the Vall d'Hebron Institute of Research (VHIR) with the goal of bringing our work on the interpretation of genetic variants closer to clinical contexts. Since then, our group has focused on developing computational methods to understand the functional impact of genetic variation in human disease. Our work has been evaluated through community challenges such as the Critical Assessment of Genome Interpretation (CAGI).