

Extensive experience and publication record in the field of molecular biology and biochemistry, with special emphasis on nervous system development in mammals. She leads the research activity of a group focusing on the human disorders xeroderma pigmentosum (XP), trichothiodystrophy (TTD) and Cockayne Syndrome (CS), which result from defects in nucleotide excision repair (NER), a versatile system that removes a wide range of lesions, including UV photoproducts. By using the next generation sequencing approaches, the group has identified novel causative genes for the NER-deficient disorders as well as genes whose transcription is deregulated in primary skin cells isolated from XP or TTD patients. This analysis allowed the identification of alterations in the extracellular matrix of TTD cells that could explain some of TTD clinical symptoms and to define new functions of the repair/transcription complex TFIIH in transcription. The use of proteomic approaches allowed the identification of novel signalling pathways involving the NER proteins and whose alterations may partially explain the clinical features of XP, TTD and CS diseases.