Research activity

She has long lasting experience in the field of human genetics with a special focus on the molecular and functional characterization of defects responsible for rare diseases. In particular, she has been involved in the definition of the primary alteration in a large sample of families with members affected by hereditary disorders defective in the DNA repair process "nucleotide excision repair", namely xeroderma pigmentosum (XP), trichothiodystrophy (TTD) and Cockayne syndrome (CS). She identified the mutations in the *ERCC2/XPD* gene underlying different pathological phenotypes (TTD, XP, combined XP/CS) and she contributed to clarify the functional role of the XPD subunit of the DNA repair/transcription complex TFIIH. She has been also involved in the characterization of the molecular defects associated to the non-photosensitive form of TTD, for which she recently identified the new causative genes *GTF2E2* and *TARS1*. Her present activity is aimed at clarifying the genetic and molecular basis of still unsolved TTD or XP cases by means of next generation sequencing technologies.

Her research activity is documented by 32 full-papers in indexed journals.