

CICCONE ROBERTO - RESEARCH ACTIVITY:

Rare genomic variants like SNVs (single nucleotide variants) and CNVs (chromosomal deletions and amplifications) are often associated with different genetic diseases. Some of these variants have a highly negative phenotypic impact and cause genetic diseases. Other variants may be not sufficient to cause a disease but represent predisposing factors for some conditions.

My research activity is focused on the identification of new genomic alterations associated with different diseases like neurodevelopmental disorders, autism, epilepsy and congenital malformations.

The identification of such variants is mainly based on the DNA analysis of subjects affected by the abovementioned conditions by using the state-of-the-art techniques for genome investigations (i.e.: chromosomal microarrays and Next Generations Sequencing).