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The role of rare and non-coding variants in the genetic predisposition to disease and drug response

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The recent explosion of high-throughput experimental methods has provided a large quantity of genetic data, improving our understanding of genetic predisposition to disease and drug response. In the past, the attention of the geneticists was focused on common variants located in coding regions, but gene-candidate and genome-wide studies demonstrated that common and coding variants explain only a slight percentage of complex phenotypes. The next-generation sequencing (NGS) technologies and the development of bioinformatics opened new routes to understand the complex architecture of human genome. In particular, recent studies have highlighted a relevant role of rare and non-coding variation in determining the expression of complex phenotype, such as disease or drug responsiveness.

Our current studies have confirmed these data. In particular, we can provide two important examples. Our investigation of human pharmacogenetic variation related to anti-hypertensive drug response demonstrated that the rare variants explain the 81% of the functionality of genes related to anti-hypertensive therapies, suggesting that a reliable pharmacogenetic test for hypertension should be based on NGS analysis. Our second example is about the transthyretin-related amyloidosis (ATTR), a monogenetic disease with a complex phenotypic variation. The pathogenesis of ATTR is due to coding mutations in *TTR* gene. However, these mutations do not explain the ATTR phenotype. Our data highlighted that non-coding variants in *TTR* gene may have an important role in gene function, explaining ATTR phenotype.

In conclusion, investigation of rare and non-coding variants may have a fundamental role to move human genetics from the bench to the bedside.

Biography

Renato Polimanti is 28 years old-candidate in the third year Ph.D. program at the University of Rome "Tor Vergata". He is author of 24 papers in Pubmed, 8 submitted articles and 1 in press book chapter. He has 151 total citations, 9 h-index, and 62.119 total impact factor. He participated at 27 international and national scientific conferences, in which he was author or co-author of 52 contributions. During his career, He won different academic awards for his scientific work and he served as associated editor, editorial board member, and reviewer of several international journals.

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