Translational research: molecular diagnostics and therapeutics

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The topic of research falls within the areas of Clinical Biochemistry and Clinical Molecular Biology. It aims to analyse the molecular basis of hereditary genetic diseases, to identify the pathogenetic mechanisms that can be targets of new personalized therapies and to propose innovative molecular diagnostics.

RESEARCH LINES
1. Study of the molecular basis and genotype-phenotype correlation in a variety of hereditary genetic diseases of cardiological relevance, such as cardiomyopathies (hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic cardiomyopathy, etc), cardiac channelopathies (long QT syndrome, Brugada syndrome, etc.), as well as numerous hereditary metabolic diseases of paediatric interest (organic aciduria, beta-oxidation defects, urea cycle defects and, globally, rare diseases of the intermediate metabolism).

2. Study of the functional effects of variants of unknown significance (VUS), by means of mRNA analysis, immunochemistry, pach-clamp techniques (also with the collaboration of international researchers).

3. Sudden death in athletes: analysis of genetic risk factors

4. Evaluation of biochemical-clinical and genetic parameters related to athletes wellness and performance.

1. The Hidden Fragility in the Heart of the Athletes: A Review of Genetic Biomarkers.
Barretta F, Mirra B, Monda E, Caiazza M, Lombardo B, Tinto N, Scudiero O, Frisso G, Mazzaccara C. Int J Mol Sci. 2020 Sep 12;21(18):6682. doi: 10.3390/ijms21186682. PMID: 32932687

2. Unexplained sudden cardiac arrest in children: clinical and genetic characteristics of survivors.
Monda E, Sarubbi B, Russo MG, Caiazza M, Mazzaccara C, Magrelli J, Rubino M, Esposito A, Perna A, Passariello A, Bossone E, Romeo E, Colonna D, Esposito MV, D'Argenio V, Salvatore F, Pacileo G, Crotti L, Frisso G, Limongelli G. Eur J Prev Cardiol. 2020 Jul 28:2047487320940863. doi: 10.1177/2047487320940863. Online ahead of print. PMID: 33611550

3. Yield and clinical significance of genetic screening in elite and amateur athletes.
Limongelli G, Nunziato M, D'Argenio V, Esposito MV, Monda E, Mazzaccara C, Caiazza M, D'Aponte A, D'Andrea A, Bossone E, Maggio FD, Buono P, Pica PW, Capua L, Penco M, Romano S, Paolo FD, Pelliccia A, Frisso G, Salvatore F. Eur J Prev Cardiol. 2020 Jul 2:2047487320934265. doi: 10.1177/2047487320934265. Online ahead of print. PMID: 32615795

4. Functional Studies and In Silico Analyses to Evaluate Non-Coding Variants in Inherited Cardiomyopathies.
Frisso G, Detta N, Coppola P, Mazzaccara C, Pricolo MR, D'Onofrio A, Limongelli G, Calabrò R, Salvatore F. Int J Mol Sci. 2016 Nov 10;17(11):1883. doi: 10.3390/ijms17111883. PMID: 27834932

5. Child neurology: Recurrent rhabdomyolysis due to a fatty acid oxidation disorder.
Terrone G, Ruoppolo M, Brunetti-Pierri N, Cozzolino C, Scolamiero E, Parenti G, Romano A, Andria G, Salvatore F, Frisso G. Neurology. 2014 Jan 7;82(1):e1-4. doi: 10.1212/01.wnl.0000438217.31437.72. PMID: 24379101