Personal data

Giulia Frisso
Born: Naples, Italy
Present Position: Associate Professor of Technical Science of Medical laboratory (SSD MED/46), University of Naples Federico II
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Education and training

1991: Degree in Medicine and Surgery with summa cum laude, University of Napoli Federico II
1992: Awarded the qualification to practize as professional medical;

1992-1996: Fellow at the Dipartimento di Biochimica e Biotecnologie Mediche, University of Naples "Federico II";
1996: Specialization in Biochemistry and Clinical Chemistry, summa cum laude, University of Napoli Federico II

2000: PhD in Biotechnology, University of Napoli Federico II

2000-2002: Post-doctoral researcher, Department of Biochemistry and Medical Biotechnologies, University of Napoli Federico II

2002-2011: Researcher of Clinical Biochemistry (SSD BIO/12), University of Napoli Federico II

2009 until now: Medical Director of the "Development of innovative technologies for the laboratory diagnosis of congenital heart and neuromuscular disease " service sector of the "Azienda Ospedaliera Universitaria Federico II";

2011 until now: Associate Professor of Technical Science of Medical laboratory (SSD MED/46), University of Napoli Federico II

Teaching experience

2002 to date: Clinical Biochemistry and Clinical Molecular Biology, Biotechnological Sciences
2006-2011: Clinical Biochemistry and Clinical Molecular Biology, Science in Physiotherapy
2007 to date: Clinical Biochemistry and Clinical Molecular Biology, Human Nutrition Sciences,
2012 to date: Laboratory Medicine, Medicine and Surgery,
2012 to date: Clinical Molecular Biology, Biomedical Laboratory Technician,
2005-to date: Invited lectures on the field of laboratory medicine for Clinical Biochemistry, Cardiac Surgery and Psychiatry Schools of Specialization
2002 to date: ECM (Continuous Education in Medicine) events in the field of Laboratory Medicine

2005: teacher and supervisor of Master BIOMEDIT in Advanced Biotechnologies for Molecular Diagnosis and Therapy

Main Research Grants from

2004-2007: MIUR-PRIN "Degenerazione maculare senile: aspetti clinico-epidemiologici e biochimico-genetici".

2008-2011: MIUR-PRIN "Identificazione precoce di marcatori genetici predittivi di patologie cardiache potenzialmente letali in atleti agonisti".

Editorial Activity
Member of the Editorial Board of Cardiogenetics (Italy) from 2011

Research activity
Research fields: development of diagnostic methods based on recombinant DNA techniques, in the field of hereditary disease, i.e.; neuromuscular disorders (Duchenne/Becker muscular dystrophy, DMD/BMD); Pyruvate kinase-deficiency (PK); Age-related Macular Degeneration (AMD); Cardiomyopathies and Channellopathies (Hypertrophic Cardiomyopathy, HCM; Long QT syndrome, LQTS), Inherited Metabolic Diseases (mainly beta-oxidation disorders and remethylation defects); Familial breast and ovarian cancer (FBOC). Diagnostic applications of high-throughput DNA sequencing.

1. Duchenne/Becker Muscular Dystrophy (DMD/BMD). Genetic diagnosis of DMD/BMD by searching for mutations in the DMD gene, which resulted in the identification of a novel deletions. Development of new methods for identification of carrier females, allowing the identification of macrodeletions, macroduplications, small insertions and small deletions of the DMD gene. Development of a quick method for linkage analysis able to unravel recombination events in the Dystrophin gene which could be seriously misleading in the molecular diagnosis.
2. Pyruvate Kinase Deficit (PKD). Search for known and unknown mutations in the gene encoding for reticulocyte pyruvate kynase (PK-RL) in patients affected by hereditary haemolytic anaemia caused by pyruvate kinase deficiency. Mutations effect on the protein structure has been studied by crystallographic analysis.
3. Age-Related Macular Degeneration (AMD). Study of genetic loci associated to AMD, confirming in the Italian population the association between AMD and the p.402Y>H (c.1277T>C) polymorphism of Complement Factor H gene (CFH). Creation and management of a Biological Samples Bank (cellular pellets, DNA, serum and plasma) of both affected AMD patients and age and sex-matched controls, developing a register of affected individuals in Italy, from which it will be possible to get epidemiological data (prevalence, age of onset, familiarity, penetrance), still lacking in our Country.
4. Hypertrophic cardiomyopathy (HCM). Genetic screening of an Italian paediatric population by searching for mutations in the sarcomeric genes, demonstrating an even greater genetic heterogeneity with respect to that already known. Application of Next generation sequencing to genetic diagnosis of HCM. Study of functional effects of the unknown mutations by means RNA and protein analysis
5. Cardiac channelopathies. Analysis of spectrum of cardiac channel mutations in unrelated Southern Italy patients affected by LQTS or Brugada syndrome. Study of functional effects of the unknown mutations by means immunochemistry and voltage-clamp techniques.

6. Inherited Metabolic Diseases. Implementation of genetic testing for the diagnosis of numerous inherited metabolic diseases, with the development of flow charts which enable the differential diagnosis between overlapping phenotypes.
7. Familial breast and ovarian cancer (FBOC). Application of Next-generation sequencing as reliable procedure for the routine molecular screening of the BRCA1/2 genes.

Publications
Over 30 original articles and three reviews have been published in peer-reviewed journals. Over 50 communications to national and international scientific conferences, also as Invited Speaker.

Editorial Activity
Member of the Editorial Board of Cardiogenetics (Italy) from 2011