

Curriculum vitae

Prof. Cecilia Gelfi

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Professor of Clinical Biochemistry and Molecular Biology
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Research activity

Muscle proteome signature under physiological paraphysiological and pathological conditions.

Brief Outline of research accomplishments

- 1) Active participation in the development of isoelectric focusing in immobilized pH gradients.
- 2) Development of a novel thermal theory for predicting with accuracy the real temperature inside the capillary during an electrophoretic run. This has allowed setting up of a novel, technique: separation of DNA fragments, carrying a point mutation, for the screening of genetic diseases. Separation of PCR amplified fragments carrying a point mutation cannot be carried out in conventional electrophoresis; the mutants can only be resolved in an electrophoretic run in the presence of a thermal gradient along the separation axis. Since it was impossible to obtain in CZE a thermal gradient in space, we have developed thermal gradients in time.
- 3) Active participation at the improvement of methods related to DNA screening particularly for point mutation detection for the diagnosis of genetic defects
- 4) Development of proteomic studies on skeletal muscle taking advantage to the knowledge on the separation science of macromolecules. In a first attempt the interest was focalized into the systematic identification of protein in a muscle tissue. A large data base of proteins localized in a high resolution 2D maps and identified by mass spectrometry has been constructed.
- 5) At present, new methodologies based on liquid chromatography and mass spectrometry are under development for the differential quantitation of high MW muscle protein, such as myosin heavy chain, titin, nebulin etc. that play a crucial role in the muscle function, but, so far, poorly characterized.
- 6) New projects are in progress involving differential proteome assessment applied to specific physiological and physiopathological problems (i.e. hypoxia adaptation, muscle impairments linked to genetic disease, effect of micro gravity, aging etc.).

Selection of Publications 1997-2009

- 1) Cremonesi, L., Firpo, S., Ferrari, M., Righetti, P.G., Gelfi, C., Double- gradient DGGE for optimized detection of DNA point mutations. *Biotechniques*, (1997) 326-330.
- 2) Cossu, G., Angius, T., Gelfi, C., Righetti, P.G.: Rapid Rh D/d genotyping by quantitative PCR and capillary zone electrophoresis: *Electrophoresis*, 17, (1996), 1911-1915.
- 3) Stoyanov, A. V., Gelfi, C. and Righetti, P.G.: Capillary zone electrophoresis of oligonucleotides in isoelectric buffers and against a stationary pH gradient. *Electrophoresis* 18 (1997) 717-723.

- 4) Gelfi, C., Righetti, P.G., Travi, M., Fattore, S.: Temperature programmed capillary electrophoresis for the analysis of high-melting point mutants in thalassemias. *Electrophoresis* 18 (1997) 724-731.
- 5) Righetti, P.G., Conti, M., Gelfi, C.: Study of haptoglobin-hemoglobin complexes by titration curves, capillary electrophoresis and capillary isoelectric focusing. *J. Chromatogr. A.* 767 (1997) 255-262.
- 6) Stellwagen, N., Gelfi, C. and Righetti, P.G.: The free solution mobility of DNA. *Biopolymers* 42 (1997) 687-703.
- 7) Righetti, P.G., Gelfi, C., Perego, M., Stoyanov, A.V. and Bossi, A.: Capillary zone electrophoresis of oligonucleotides and peptides in isoelectric buffers: theory and methodology. *Electrophoresis* 18 (1997) 2145-2153.
- 8) Perego, M., Gelfi, C., Stoyanov, A.V. and Righetti, P.G.: Separation of oligonucleotides of identical size, but different base composition, by free zone capillary electrophoresis in strongly acidic, isoelectric buffers. *Electrophoresis* 18 (1997) 2915-2920.
- 9) Gelfi, C., Righetti, S.C., Zunino, F., Torre, G.D., Pierotti, M.A. and Righetti, P.G.: Detection of p53 point mutations by double-gradient, denaturing gradient gel electrophoresis. *Electrophoresis* 18 (1997) 2921-2927.
- 10) Carrera, P., Barbieri, A.M., Ferrari, M., Righetti, P.G., Perego, M., Gelfi, C.: Rapid detection of 21-hydroxylase deficiency mutations by allele-specific in vitro amplification and capillary zone electrophoresis. *Clin. Chem.* 43 (1997) 2121-2127.
- 11) Gelfi, C., Perego, M., Righetti, P.G., Cainarca, S., Firpo, S., Ferrari, M. and Cremonesi, L.: Rapid capillary zone electrophoresis in isoelectric histidine buffer: high resolution of the poly-T tract allelic variants in intron 8 of the CFTR gene. *Clin. Chem.* 44 (1998) 906-913.
- 12) Gelfi, C., Curcio, M., Righetti, P.G., Sebastiano, S., Citterio, A., Ahmadzadeh, H. and Dovichi, N.: Surface modification based on Si-O and Si-C sublayers and a series of N-substituted acrylamide top layers for capillary electrophoresis. *Electrophoresis* 19 (1998) 1677-1682.
- 13) Gelfi, C., Mauri, D., Perduca, M., Stellwagen, N.C. and Righetti, P.G.: Capillary zone electrophoresis of ds-DNA in isoelectric buffers: effect of adding of competing, non-amphoteric ions. *Electrophoresis* 19 (1998) 1704-1710.
- 14) Cremonesi, L., Carrera, P., Fumagalli, A., Lucchiari, S., Cardillo, E., Ferrari, M., Righetti, S.C., Zunino, F., Righetti, P.G. and Gelfi, C.: Validation of double gradient, denaturing gradient electrophoresis through multigenic retrospective analysis. *Clin. Chem.* 45 (1999) 35-40.
- 15) Carrera, P., Piatti, M., Stenirri, S., Grimaldi, L.M.E., Marchioni, E., Curcio, M., Righetti, P.G., Ferrari, M., Gelfi, C.: Genetic heterogeneity in Italian families with familial hemiplegic migraine. *Neurology* 53 (1999) 26-32.
- 16) Battistini, S., Stenirri, S., Piatti, M., Gelfi, C., Righetti, P.G., Rocchi, R., Giannini, F., Battistini, N., Guazzi, G.C., Ferrari, M. and Carrera, P.: A new CACNA1A gene mutation in acetazolamide-responsive familial hemiplegic migraine and ataxia. *Neurology* 53 (1999) 38-43

- 17) Gelfi, C.; Viganò, A., Carta, P., Manchi, P., Cossu, G. and Righetti, P.G.: screening for the β -39 mutation by capillary electrophoresis in free solution in strongly acidic, isoelectric buffers. *Electrophoresis* 21 (2000) 780-784.
- 18) Gelfi, C.; Viganò, A., Curcio, M., Righetti, P.G., Righetti, S.C., Corna, E. and Zunino, F.: Single- strand conformation polymorphism analysis by capillary zone electrophoresis in neutral pH buffer. *Electrophoresis* 21 (2000) 785-791.
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- Gelfi, C.**, De Palma, S., Cerretelli, P., Begum, S., Wait, R.: Two dimensional protein map of human vastus lateralis muscle. *Electrophoresis* 2003, 24, 286-295.
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- 124) A Pontoglio, A.Viganò, R. Sebastiano, A. Citterio, L. Maragnoli, P.G. Righetti and **C.Gelfi**. Peptide and protein separations by capillary electrophoresis in presence of mono- and di-quaternarized diamines, *Electrophoresis*, 2004, 25,1065-1070.
- 125) Righetti PG, **Gelfi C**, Sebastiano R, Citterio A. Surfing silica surfaces superciously *J Chromatogr A*. 2004, 1053:15-26.
- 126) D Capitanio, A. Viganò, E. Ricci, R. Wait, P. Cerretelli, **C. Gelfi**. Comparison of protein expression in human deltoideus and vastus lateralis muscles using two-dimensional electrophoresis., *Proteomics*, 2005 , 5, 2577-86 .
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- 128) De Palma S, Morandi L, Mariani E, Begum S, Cerretelli P, Wait R, **Gelfi C**, Begum S, Cerretelli P, Wait R, Gelfi C.
Proteomic investigation of the molecular pathophysiology of dysferlinopathy. *Proteomics*. 2006, 6:379-85.
- 129) **Gelfi, C.**, Viganò, A., Ripamonti, M., Pontoglio, A., Begum, S, Pellegrino, MA., Grassi, B., Bottinelli, R., Wait, R., Cerretelli, P. The human muscle proteome in aging, *J. of Proteome Research*, 2006, 5, 1344-1353.

- 130) Celegato B, Capitanio D, Pescatori M, Romualdi C, Pacchioni B, Campanaro S, Viganò A, Wait R, Colantoni L, Ricci E, Lanfranchi G and **Gelfi C**. Parallel protein and transcript profiles of FSHD patient muscles correlate to the D4Z4 arrangement and reveal a common impairment of slow to fast fibre differentiation and a general deregulation of MyoD-dependent genes. *Proteomics*, 2006, 6, 5303-5321.
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