

Claudio Graziano

Curriculum Vitae

Personal data

Date of birth: 12 April, 1972
Place of birth: Pesaro, Italy
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Current position

Consultant in Medical Genetics at Azienda Ospedaliera di Bologna Policlinico S. Orsola-Malpighi.

Education

1989-90: Student at Concord High School in Concord, New Hampshire, USA, holding an “American Field Service” scholarship.
October 1997: degree *cum laude* in Medicine and Surgery at the University of Florence with a thesis on “Molecular analysis of tumor suppressor gene TP53 carboxy-terminal region in Head and Neck Cancer” (tutor Professor Pier Luigi Mattiuz).
November 2002: degree of Specialization in Medical Genetics at the University of Florence with a thesis on “Clinical and molecular analysis in patients with Nemaline Myopathy”.

Research experiences abroad

2001: January – June: host at the Clinical Genomics Unit, *Center for Genomics and Bioinformatics* of Karolinska Institut in Stockholm.

Clinical and scientific activity

2002 – 2004: Research fellowship at the Department of Clinical Physiopathology, University of Florence.
2003: Activity of genetic counselling in the Unit of Medical Genetics (Director Prof. Giovanni Romeo) at Azienda Ospedaliera di Bologna Policlinico S. Orsola-Malpighi.
2004 – 2005: Research fellowship at the Department of Internal Medicine, Hepatology and Cardioangiology of the University of Bologna, with acknowledgement of the activity of genetic counselling in the Unit of Medical Genetics.

2006 – 2007: Researcher of Medical Genetics from 02/05/2006 to 28/02/2007 at the Department of Clinical Physiopathology, University of Florence, Unit of Medical Genetics (director Prof. Maurizio Genuardi).

Publications

- Wallgren-Pettersson C, Pelin K, Hilpela P, Donner K, Porfirio B, Graziano C, Swoboda KJ, Fardeau M, Urtizberea JA, Muntoni F, Sewry C, Dubowitz V, Iannaccone S, Minetti C, Pedemonte M, Seri M, Cusano R, Lammens M, Castagna-Sloane A, Beggs AH, Laing NG, de la Chapelle A. Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. *Neuromusc Disord* 1999; 9: 564-572
- Porfirio B, Chiarelli I, Graziano C, Mannoni A, Morrone A, Zammarchi E, Beltràn-Valero de Bernabé D, Rodriguez de Cordoba S. Alkaptonuria in Italy: polymorphic haplotype background, mutational profile, and description of four novel mutations in the homogentisate 1,2-dioxygenase gene. *J Med Genet* 2000; 37: 309-312
- Graziano C, Bertini E, Minetti C, Porfirio B. α -actin gene mutations and polymorphisms in Italian patients with Nemaline Myopathy. *Int J Molec Med* 2004; 13: 805-809
- Vannucchi AM, Grossi A, Pancrazzi A, Antonioli E, Guglielmelli P, Balestri F, Biscardi M, Bulgarelli S, Longo C, Graziano C, Gugliotta L, Bosi A. *PRV-I*, erythroid colonies and platelet Mpl are unrelated to thrombosis in essential thrombocythaemia. *British J Haem* 2004; 127: 214-219
- Graziano C, Bertini E, Porfirio B. *De novo* α -actin mutations in monozygotic twins. *Clinical Genet* 2005; 68:91-92
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- D'Amico A, Graziano C, Pacileo G, Petrini S, Nowak KJ, Boldrini R, Jacques A, Feng JJ, Porfirio B, Sewry CA, Santorelli FM, Limongelli G, Bertini E, Laing N, Marston SB. Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. *Neuromusc Disord* 2006; 16: 548-552
- Turchetti D, Razzaboni E, Zomer H, Rossi C, Ferrari S, Greco D, Graziano C, Romeo G, Seri M. Psychological consequences of prenatal diagnosis in a case of familial Angelman Syndrome. *Prenat Diagn.* 2006; 26(12):1156-9
- Jaijo T, Aller E, Beneyto M, Najera C, Graziano C, Turchetti D, Seri M, Ayuso C, Baiget M, Moreno F, Morera C, Perez-Garrigues H, Millan JM. MYO7A Mutation Screening in Usher Syndrome Type I Patients from Diverse Origins. *J Med Genet* 2007; 44(3):e71
- Graziano C, D'Elia AV, Mazzanti L, Moscano F, Guidelli Guidi S, Scarano E, Turchetti D, Franzoni E, Romeo G, Damante G, Seri M. A De Novo Nonsense Mutation of PAX6 Gene in a Patient with Aniridia, Ataxia and Mental Retardation. *Am J Med Genet* 2007; 143A:1802-5

Affiliations

Registered at the Medical Association of Florence since September 1998
Affiliated to the Italian Society of Human Genetics since 1999