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Short biography

Dr. Schiaffino obtained her MD and PhD from the University of Padova Medical School, Italy, in 1991 and 1996. She then moved as a research associate to the Department of Human and Molecular Genetics, Baylor College of Medicine, Houston, USA, where she began working on Ocular Albinism type 1 with Dr. Andrea Ballabio. Back to Italy in 1995, initially as a researcher at Telethon Institute of Genetics and Medicine (TIGEM), in Milan, she was subsequently appointed as group leader and head of research unit, at Scientific Institute San Raffaele, Milan, in 2000 and 2006. Her research work both as postdoc and independent investigator is mainly focused on the molecular genetics and cell biology of pigment cells and their alterations in Ocular Albinism, as demonstrated by several publications and invited presentations on the topic.

Research interests

Dr. Schiaffino has dedicated most of her scientific career to the pathogenesis of Ocular Albinism type 1 at the molecular and cellular levels, including the isolation of the involved gene (Bassi/Schiaffino et al. *Nature Genetics* 1995); mutation identification and characterization (Schiaffino et al. *Human Molecular Genetics* 1995; Bassi et al. *Human Genetics* 2001; d'Addio et al. *Human Molecular Genetics* 2000); in vitro gene therapy studies in human melanocytes (Schiaffino et al. *Human Gene Therapy* 2002); functional characterization of the OA1 protein and its role in pigment cells (Schiaffino et al. *PNAS* 1996; Schiaffino et al. *Nature Genetics* 1999; Piccirillo et al. *Journal of Cell Science* 2006; Innamorati et al. *Pigment Cell Research* 2006; Palmisano et al. *Human Molecular Genetics* 2008; Daniele et al. *Current Biology* 2014; Falletta et al. *Pigment Cell and Melanoma Research* 2014). Her current research interests regard the identification of potential genetic and pharmacological targets/strategies for the treatment of the disease.