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Short biography

Benoit Arveiler was trained in Pharmacy (Strasbourg, 1979-1984). He then did a Master in Cellular and Molecular Biology (Strasbourg, 1984-1985) and a PhD in Molecular Genetics (Strasbourg, 1985-1988) in Pr. Jean-Louis Mandel's laboratory. After a post-doctoral training in Edinburgh (1989-1992), he was appointed as an Assistant at University of Bordeaux and University Hospital of Bordeaux in 1992, where he then became a Lecturer in 1994, and then a Professor in Medical Genetics in 1998. In 1992 he established the Molecular Genetics Laboratory at University Hospital of Bordeaux, a clinical laboratory of which he still is the director. At the same time he established a research laboratory at Bordeaux University of which he has been the head until 2010.

He is also strongly involved in the organization of a network of French molecular genetics laboratories and is since 2013 the President of the French Association of Molecular Genetics Practitioners. He is also very much involved in the implementation of the French Rare Disease Plan and the French Genomic Medicine Plan.

In the field of albinism, he participated in the establishment of the Albino Days (multidisciplinary consultations) at Bordeaux University Hospital in 2014. He has co-organized the 1st European Days of Albinism in 2012, and has been a member of the scientific committees of the 2nd and 3rd EDAs.

He is the coauthor of more than 120 publications, including 13 on albinism.

Research Interests

Benoit Arveiler has always been working in the field of molecular genetics. Most of his involvement over the last fifteen years was on albinism, a period during which he gradually established the thorough analysis of the albinism genes for diagnosis purposes. This nowadays includes the systematic search for point mutations and gene rearrangements in the 19 genes implicated in ocular, oculocutaneous and syndromic forms of this condition. Considering that about 20-25% of patients remain without a molecular diagnosis, his present work focusses on the search for new albinism genes by exome and candidate gene approaches. A search for regulatory and intronic mutations has also been undertaken.