

Short bio

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I started medical studies at the University of Bochum in Germany, but participated also in exchange programs with French universities (Strasbourg, Paris, Montpellier). My interest in immunology dates back to my years in medical school when I did an experimental work on the complement system for my MD thesis. During an internship in the Department for Immunology (Pr Jean-Louis Pasquali) in Strasbourg, France, I had the chance to gain first clinical insights into the complexity of immunological disorders in adults. Interested in the pathophysiology of the immune system, I interrupted my residency in Paediatrics in order to perform basic research in the INSERM laboratory of Alain Fischer in Paris, France. Under the supervision of Jean-Pierre de Villartay, I explored the molecular basis of yet genetically undefined forms of T-B⁻NK⁺ SCID patients with a special interest in V(D)J recombination/DNA repair. I was fortunate enough to identify the gene coding for Artemis, a key player in V(D)J recombination/DNA repair, and to investigate patients with other V(D)J recombination defects like Rag1/2 or DNA Ligase IV.

After my PhD thesis, I continued my training in paediatrics. I had the opportunity to gain further clinical experiences in the field of primary immunodeficiencies by working in several specialized centres for Paediatric Immunology and Haematology throughout Europe, and I owe a lot to my mentors: Professor Alain Fischer in Paris, Professor Gigi Notarangelo with whom I worked while he was still in Brescia, Italy, and Professor Ulrich Göbel in Düsseldorf, Germany.

Since 2005, I am working as a paediatrician in the Unit for Paediatric Immunology, Haematology and Rheumatology at Necker-Enfants Malades University Hospital in Paris, the national reference centre in France for patients with rare diseases of the immune system. In the clinical unit, I am coordinating diagnostic and therapeutic strategies for the patients, as well as their day-to-day care. I have a longstanding and solid clinical experience in allogeneic HSCT, as we are performing up to 50 HSCT for inborn errors per year in our unit. We are also participating in gene therapy trials.

Particularly interested in primary HLH, I launched a national prospective treatment study in France (Alemtuzumab as first line treatment in Familial Lymphohistiocytosis). In this respect, I am active member of the Histiocyte Society and the HLH steering committee. I am also involved in improving diagnostic and therapeutic approaches for patients with primary immunodeficiency through national and international collaborative studies.

My current position as Professor for Paediatrics at Université Paris Cité gives me the unique opportunity to work at the interface between basic and clinical research. I have the privilege to conduct my research activity in the group “Genome dynamics in the immune system” within the Imagine Institute, a research and innovative healthcare institute located on the campus of the Necker-Enfants Malades Hospital. Imagine Institute is bringing together researchers, doctors and patients, with a common goal: to cure genetic diseases (www.institutimagine.org).

I am active member of ESID since 2005 and also active member of the IEWP. I have been the chair of the Clinical WP (2012-2016) and the Education WP (2018-2022), and I remain involved in educational activities. It's a real pleasure to continue working closely together with Clara Franco Jarava, Antonio Marzollo, and Boris Karanovic, as I am passionate about teaching. Currently, I am at the end of my term as President of the French Society for Paediatric Haematology and Immunology (SHIP). Since 2022, I am directing the CEREDIH, the French National Reference Centre for Hereditary Immune Deficiencies.

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