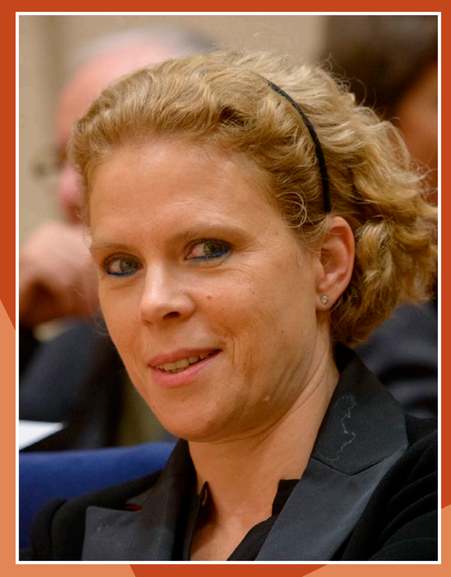
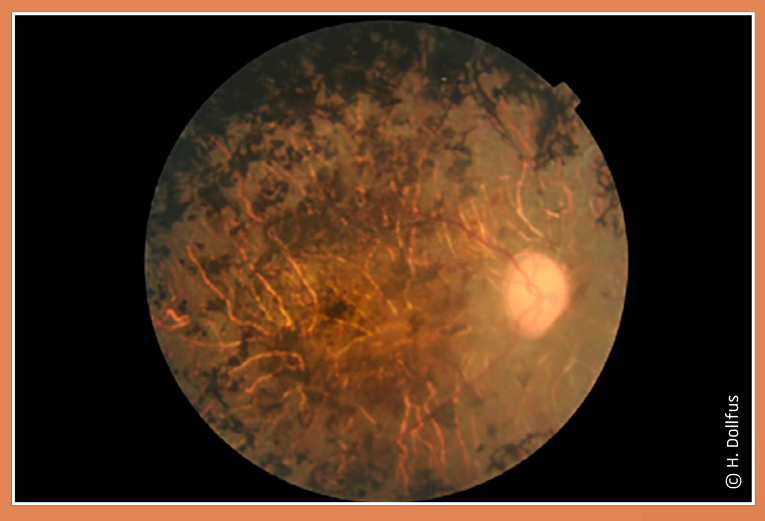
Hélène Dollfus
USIAS Paul Ehrlich Chair (2024-2026)

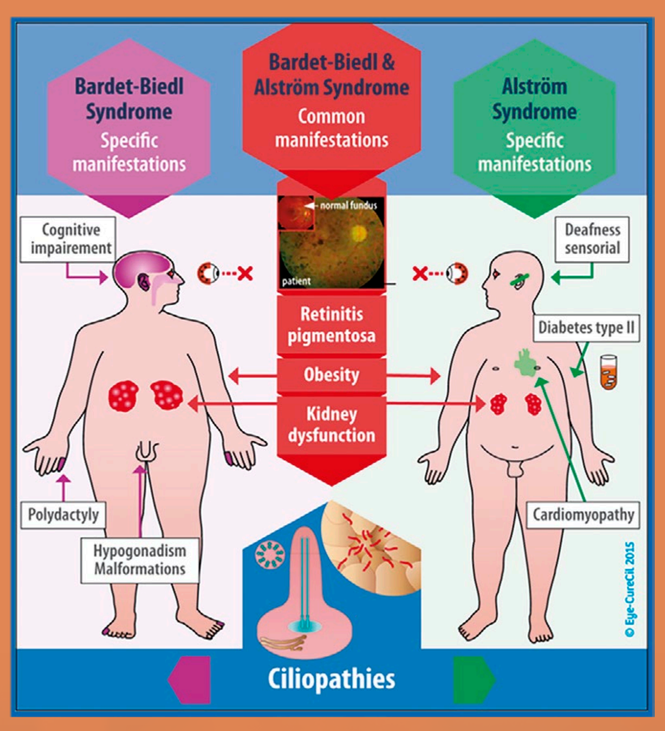


Hélène Dollfus is professor of medical genetics at the University of Strasbourg and a consultant at the University Hospital of Strasbourg (HUS). Based at the Strasbourg Biomedicine Research Centre (CRBS) she heads the HUS medical genetics department and the Laboratory of Medical Genetics (LGM). Clinically and scientifically, her work focuses on rare and ultra-rare genetic syndromes, particularly those affecting the eye.

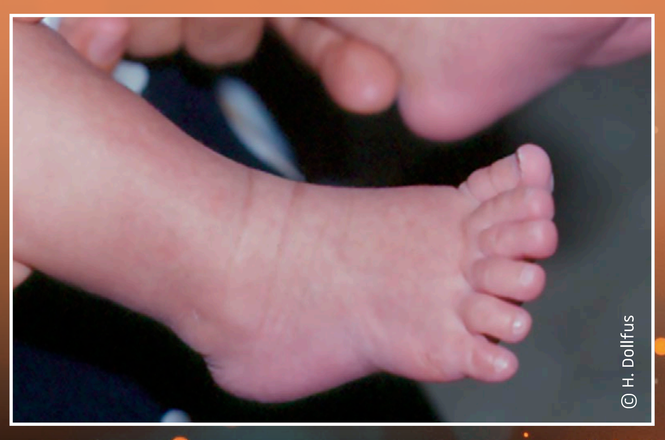


Severe retinitis pigmentosa causing loss of vision, related to the BBS12 gene.

By definition, any rare disease affects only a small number of people. However, with over 6,000 types of rare diseases in existence, their global burden is high, with an estimated prevalence of up to 6% of the world population. They are typically chronic, progressive, and very debilitating – and most lack approved treatments. Around 80% of rare diseases have a genetic cause.



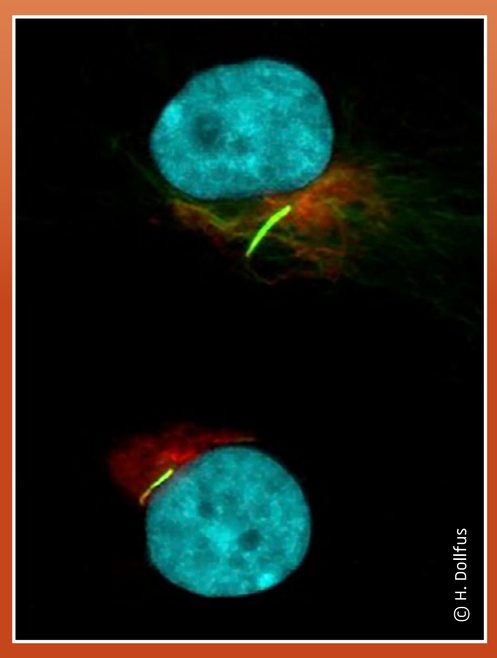
"As a young clinician working on rare genetic eye diseases, I once came across two families with children affected by the ultra-rare Bardet-Biedl syndrome on the same day. This serious and untreatable condition leads to early-onset blindness and a range of other debilitating symptoms related to multiple organ damage. At the time, this syndrome was a mystery, and there was hardly any research into it, because it affected so few people. I was very moved by the desperation of the families. And, in addition, I was intrigued. My scientific curiosity was triggered, and has never waned since."



◀ Polydactyly (extra fingers or toes) is a characteristic feature of Bardet-Biedl syndrome.

Rare genetic diseases not only represent a serious public health issue worldwide, but also an exciting field of research and a great opportunity for scientific discovery, because they combine so many facets of medicine. They offer a unique window into basic genetic processes, which are very hard to observe or even detect in other ways.

The work of Professor Dollfus focuses on the dysfunction of the primary cilium (ciliopathies), an antenna-like organelle that extends from the cell and can sense the surrounding microenvironment. Defects in cilia function underlie a wide range of – diverse but related – developmental or degenerative human diseases. Her research has led to the identification of over a dozen of genes responsible for ultra-rare syndromes including ciliopathies, and has also revealed a very broad clinical spectrum, providing an example of phenotypic variability.



Human fibroblasts in culture, with primary cilia and the ARL13B ciliary antibody.

"Working on rare diseases may serve much larger populations of patients with more common diseases, as this research can reveal new insights at the basic level, and pave the way for the development of pharmacological and genetic therapies for treatment that may, at some point, also be applied in other areas."

Hélène Dollfus initiated the Institute of Medical Genetics of Alsace (IGMA), and is coordinator of the national health network of rare sensorial diseases (SENSGENE) and of ERN-EYE (European Reference Network for Rare Eye Diseases) that brings together over 60 hospitals in almost all EU Member States.

"My main goal is to help patients and their families – through new knowledge, but also through better use of existing knowledge and improved care. We will not leave them on their own."

The Paul Ehrlich Chair in the life sciences was created in 2022 for Strasbourg-based researchers who have made an exceptional contribution to their field. The Chair is named in honour of Paul Ehrlich (1854-1915), a German physician and scientist who studied in Strasbourg and is widely recognized for his research on haematology, immunology and pharmacology. Known as the father of chemotherapy, he was awarded the 1908 Nobel Prize in Physiology or Medicine for his contributions to immunology.

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