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With an estimated prevalence of 12 in 100,000, cystic fibrosis is a rare disease. It has the distinction, however, of being the most frequent of the monogenic diseases that are potentially serious from childhood in populations of European origin. A hereditary illness with autosomal recessive transmission, it only affects individuals with homozygous mutations: that is, who have inherited two mutated versions of the same gene, one from the father and the other from the mother. The disease is chronic, progressive, and most often expressed very early in infancy or even from birth. It has both respiratory and digestive symptoms, but in most cases it is the severity of respiratory dysfunction that determines the course of the disease's threat to the individual's life.

In this book, Georges Travert narrates the history of the medical advances and discoveries that have contributed over time to the understanding of this complex genetic disease and the most promising treatments for it. The first accounts of the disease—long unknown to physicians because of its rarity—likely date to the early 17th century, when it was associated with witchcraft: 'Woe to the child who tastes salty from a kiss on the brow, for he is cursed and soon will die.' But it was not until the first half of the 20th century that cystic fibrosis became a distinct pathological entity, with clinical descriptions associating specific respiratory and digestive abnormalities in children with this sign first observed centuries earlier—a reflection, it turned out, of an excessive concentration of chlorine and sodium in sweat. In 1989, with the identification of a gene and its functions, followed by the identification of abnormalities caused by a particular mutation in it, researchers were able to get a complete picture of the pathophysiological characteristics of cystic fibrosis and to begin to devise targeted therapeutic strategies.

The story of this medical adventure is also a story of scientific collaborations, debates, and disagreements between competing groups of researchers and practitioners, and the role played by families and associations in the organizations that raised the necessary funds for research and the creation of patient registers. The book is also a plea for universal neonatal screening for cystic fibrosis, an area in which Travert has been one of the main players in France. While in the past this practice has often been met with scepticism from a part of the medical profession, today it is an essential component in the diagnosis of the disease and the effective care and treatment of those affected by it.

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