

GAGNA A. & CH. VAN HECK PRIZE FOR INCURABLE DISEASES – 2021

LAUREATE

The "**GAGNA A. & CH. VAN HECK PRIZE FOR INCURABLE DISEASES - 2021**" has been awarded to **Professor Guillaume CANAUD**, from the Necker Hospital, University of Paris (France), for his contributions to the therapy of a spectrum of overgrowth syndromes characterized by the presence of genetic alterations in a gene called PIK3CA and grouped under the acronym of PROS (PIK3CA-related overgrowth syndrome).

From the clinical standpoint, these conditions are highly diverse, including malformations of the vascular tree, of the adipose tissue and of the bone. From the genetic standpoint, they are caused by activating mutations in a gene called PIK3CA, which codes for a molecule (PI3K) regulating cell growth. No therapy was available for these patients, who often required mutilating surgery.

Based on his knowledge of the molecular mechanisms underlying the disease, Dr. Canaud put forward the idea that inhibitors of the PI3K complex, which are used for certain types of cancer patients, could be employed for these patients. The results of a pilot trial on 19 patients indicated significant clinical benefit to all patients, with dramatic improvement of symptoms.

This success story underlines the importance of doctors-scientists who build on the knowledge of how a disease develops to devise novel therapeutic strategies.

