COSY
Cure Overgrowth Syndromes

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PRESS RELEASE
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COSY's team is pleased to present you our website http://www.rhu-cosy.com created to promote our work and progress.

WEB SITE
RHU-COSY.COM

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+ What is RHU-COSY?

COSY (Cure Overgrowth Syndromes) is 5-year project funded by the French National Research Agency (Agence Nationale de la Recherche - ANR) through its 'Investment for the Future' Program (Programmes d'Investissement d'Avenir - PIA), under the 'University-Hospital Research in Health' action (Recherche Hospitalo-Universitaire - RHU). The project comprises partners, including scientists, geneticists, physicians, and industrial partners, all engaged in finding new therapeutic approaches to treat Overgrowth Syndromes (OS) and improving patient's medical care.

Our main goal is to transform the outcome and the medical care of patients with OS. Therefore, we aim to better understand overgrowth physiopathology, create expert centers, develop new imaging software, envision new therapeutics for precision medicine, and improve patients' socialization.

+ What is the basis of the project?

The project is based on recent work that the scientific coordinator, Pr Guillaume Canaud, published in the journal Nature. In this study, Pr Canaud and his Team demonstrated that it was possible to create experimental models of overgrowth syndromes, and to identify and directly reposition drugs to clinical application.

+ What are overgrowth syndromes?

Overgrowth syndromes are defined as a group of rare genetic syndromes characterized by malformations and tissue proliferation caused by somatic mutations that usually occur during embryonic development, in other words, after fertilization. These mutations can affect any type of tissue as well as different mTOR genes of which the most well-known is PIK3CA. Although these syndromes are considered as rare disorders, their exact prevalence is yet unknown. It is likely to be underestimated owing to the variability of the clinical presentation and the broad phenotypic spectrum of the diseases.

+ **For any further information on RHU-COSY,** please send an email to : contact@rhu-cosy.com

Interview of Pr Guillaume Canaud
Coordinator of the RHU-COSY

● What is your role in this project?

‘ I am the scientific coordinator of this consortium which brings together over a dozen of academic partners, hospitals, private companies, and a Foundation. This sort of research consortium is unique and a specific French feature.‘

What are your motivations?

‘ It’s a taxing question! I’ve always been drawn to science, understanding how a disease works, but also very excited by novelties. Moreover, I am very sympathetic to the issue of disability in this type of pathology, the consequences are very significant. In addition to the symptoms of the disease such as pain or bleeding, repeated hospitalizations and surgeries, the social repercussion is major with far too often children and adults discriminated because of physical disabilities and are thus on the margins of society. Lastly, when you see the enthusiasm of the different teams involved (research laboratory, paramedical and medical staff…) your motivation is increased tenfold! ‘

Which disease are your work focus on?

‘ We are currently working on a group of pathologies responsible for overgrowth syndromes. Of course, we have a particular interest in mutations of the PIK3CA gene involved among other things in CLOVES syndrome or venous and lymphatic vascular abnormalities, but also in genes called AKT1 (responsible for Protea syndrome) and his cousins AKT2, AKT3 and mTOR. This project is also expected to identify new genes and possibly new therapeutic avenues. Indeed, this is one of the major challenges of the project, which involves repositioning treatments developed for other pathologies that could be beneficial in these syndromes, as we have already demonstrated.‘

How do you intend to study it?

‘ Thanks to this funding, we have a unique opportunity to bring altogether different categories of research (clinical, fundamental, genetic, radiological, computer...) around the same type of pathology. In my opinion, the key element in this kind of project is to have patients and basic research on the same place (new mouse models, new genes identified…). That’s what we came to create on the Necker campus.‘

What are your hopes and goals for the future?

‘ This is a very promising project since it will provide a better understanding of these diseases and help to identify new markers of the activity of these diseases, to identify new therapeutic targets, to find new genes involved in these syndromes, to improve the integration of patients into society and finally to make these pathologies known to the medical community in order to improve their cares.‘