CHOPS Rare Diseases Foundation launches investigation into biological mechanisms of the rare syndrome

Three international contracts signed

From left to right, the researchers Kosuke Izumi, Ilaria Parenti and Jun Young Sonn

ITALY, December 6, 2024 /EINPresswire.com/ -- "We have signed 3 contracts and are now ready to see basic research investigating the as-yet-unknown biological mechanisms that characterise CHOPS syndrome come to life. These are important contracts with authoritative institutions. Kosuke Izumi, Ilaria Parenti and Jun Young Sonn. They are the professionals who mark a crucial moment in the Foundation's work". Manuela Mallamaci, president of CHOPS Rare Diseases Foundation and the mother of little Mario, announced this.

The CHOPS Foundation is the world's first and only non-profit organisation to research a cure for this rare, multi-organ syndrome.

CHOPS syndrome, an acronym representing its symptoms - cognitive impairment and coarse facies, heart defects, obesity, pulmonary involvement, and skeletal dysplasia and short stature - currently has only 34 documented cases worldwide, including that of little Mario in Italy.

Kosuke Izumi, from the University of Texas Southwestern Medical Center in Dallas, first in line for the $100,000 + $100,000 grant. Ilaria Parenti, from the Institute of Human Genetics at the Essen University Hospital of Duisburg University in Germany, and Jun Young Sonn, from Baylor College of Medicine in Houston, Texas, tied for the $20,000 grant. These are the 3 researchers who will start up the first projects that have been announced and awarded, following the evaluation of the Foundation medical-scientific advisory board.

Lawyer Marco Del Pinto, founder of Del Pinto e Associati and partner of the Foundation: "Our firm has been assisting the Foundation pro bono since its establishment, and it is with great

satisfaction that it has also provided legal support for the negotiation and drafting of the last 3 research contracts signed with internationally prestigious institutions and researchers. These agreements represent a crucial step in consolidating strategic collaborations aimed at investigating and tackling a syndrome that is as rare as it is complex, as well as a fundamental milestone for the Foundation, guaranteeing a solid legal basis to pursue its mission of research and innovation. Thanks to our work, the Foundation's commitment, donations, and scientific expertise, we will soon arrive at helpful knowledge that will bring us closer to discovering a cure".

"We are delighted with this achievement, which now also commits the Foundation to basic research. Applied research has already been started. It is our intention to proceed with the two activities in parallel. We consider it strategic to ensure the effectiveness of our research. A few months ago, in fact,' explains Mallamaci 'the company Unravel Bioscience provided us with a list of very promising drugs to be tested that could alleviate one or more symptoms of CHOPS and improve patients' quality of life. We are now developing the next phase. We are going to fund a project to test the drugs in the lab on cell lines obtained from the CHOPS samples from different continents. In vivo testing will also be necessary. We also involved another group of 8 Cornelia De Lange patients. After all, it is our goal to space out research in the field of rare neurodevelopmental diseases' continues Mallamaci.

The medical-scientific advisory board, composed of Ian Krantz, CHOPS discoverer with Izumi with the first diagnosis made at the Children's Hospital of Philadelphia in 2015, Andrea Ballabio, Valentina Massa, Ali Shilatifard, Emanuela Scarano, Katsuhiko Shirahige, Eleonora Orlandini, Sara Bozzetto, and Neil Hackett, guarantees the work of the Foundation.

"We do not intend to stop, but we need the solidarity and support of the people. The Foundation has already started projects with more than 300,000 euros raised. Complex and challenging projects. It will be crucial,' Mallamaci emphasises, 'to continue by activating the best minds on an international level to shed light on this disease. So, it will be necessary to continue raising funds. Path Along, which we are also using, is the CHOPS documentary donated and created by the director Antonio Melasi.
The Foundation is committed to promoting its work internationally and involving families in this awareness-raising process.
"We have established contacts that we consolidated last July during the CHOPS symposium, which we co-organized at the Children's Hospital of Philadelphia. We maintain an open approach focused on research and information sharing to enhance awareness of the many diseases that remain rare today," concludes Manuela Mallamaci, president of the CHOPS Rare Diseases Foundation and mother of Mario.

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