Fondazione Telethon has allocated significant funds to a research project on the West syndrome, a complex pathology that affects the nervous system, headed up by Antonello Mallamaci, a neurobiologist at SISSA of Trieste.

Antonello Mallamaci will be coordinating a research project on the West syndrome, a devastating disorder that manifests itself at an early age in infants. The study conducted by Mallamaci and his colleagues will be made possible thanks to the funding recently assigned – following a thorough evaluation on the part of the medical-scientific committee of Fondazione Telethon – to the International School for Advanced Studies (SISSA) of Trieste, and in particular to the Laboratory of Cerebral Cortex Development (CerCorDevLab), where researchers explore the molecular mechanisms that in the embryo guide the development of the cerebral
The West syndrome is characterized by spasms (often followed by intense crying fits) and by massive electroencephalographic alterations. The pathology is also characterized by the slowing down, or even the regression, of the psychomotor development: many of the children affected by it, for instance, are still unable to stay in a sitting position aged one. Spasms may disappear with treatment, yet psychomotor regression may in any case remain. The causes for the syndrome are rather diverse. It was recently discovered that one specific variant of the disorder arises due to the presence of an “excess” copy of the Foxg1 gene, which is implied in the development of the cerebral tissue and already the object of previous studies by Mallamaci and his team.

“Our project, which Telethon chose to finance, aims at retracing how a of Foxg1 overdose may trigger the pathogenic mechanism at the core of the syndrome,” explains Mallamaci. “This will help us obtain useful indications on possible points of attack on molecules in view of an effective gene therapy intervention.”

“The project will benefit from the contribution of two key partners. Yuri Bozzi of Università di Trento will take care of the behavior analysis of the animal models of the disorder we have generated - adds the neurobiologist - while Pasquale Striano of Istituto Gaslini of Genoa will provide the biological specimens and clinical profiles of the patients involved in the study.”

The total funding to be allocated by the Fondazione amounts to €260,000 and shall be assigned to two projects for the city of Trieste. Besides the project carried out by Mallamaci’s team, in fact, also the study on hereditary thrombocytopenia conducted by Anna Savoia of Università di Trieste will be financed.

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