Basic Research

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The Research

Dr. Roux is researching gene therapy in mice using *BDNF* (Brain Derived Neurotropic Factor) instead of *MECP2*. The study evaluates whether or not there can be effective treatment which bypasses MeCP2. Dr. Roux is a new Principal Investigator for Rettsyndrome.org. To read more about his research, visit http://www.rettsyndrome.org/file/18-research-files/Roux-InDepth.pdf

The Hope

The use of gene therapy in Rett syndrome aimed at improving all Rett symptoms. Based on the advances made in this area by several teams, we believe that the future results of this project will open up new therapeutic opportunities for children with Rett Syndrome.

The Answers to your Questions

What is the most exciting/hopeful aspect of this project and its possible results?

BDNF is probably the most studied factor in Rett syndrome and its therapeutic power should be obvious. However, to our, knowledge, no team has yet sought to evaluate its effectiveness in gene therapy. We hope this treatment will be able to reduce a large number of symptoms in our Rett mouse model. This will provide much needed preclinical data that will help us determine if such treatment could ever be used in patients.

Why is this work important to helping my child?

Our translational research work will allow us to move a little further towards the development of new therapeutic approaches.

What are you looking for/measuring/trying to solve in simple terms?

Since BDNF deficiency has been correlated to motor, cognitive and autonomic dysfunctions in Rett syndrome, we hope to alleviate most of these symptoms in our preclinical mouse model.

Does the knowledge gained help treat Rett or cure Rett?

Our goal is directly dedicated to curing Rett syndrome. ….

What is the timeline of your work?

Thanks to the two-year funding provided by Rettsyndrome.org, we will be able to start investigating this new gene therapy approach. This will be enough time to start investigating the safety and efficacy of this therapy and gather preclinical data. However, should this new therapy be successful, much more work will be needed before it is deemed safe for use in Rett patients

The Researcher

Dr. Roux has a long-standing interest in developmental neurophysiology. His expertise in breathing control lead him to work on Rett syndrome, and Dr. Roux was part of one of the first team describing breathing defects in a preclinical model of Rett syndrome. Following on these results, Dr. Roux proposed a pharmacological treatment able to improve breathing defects and to increase the lifespan of a Rett mouse model, which laid the groundwork for a phase IIa clinical trial under the supervision of Professor Josette Mancini (Pediatric Neurology Dept., Marseille University Hospital). Thereafter, focusing on other systems affected by MeCP2, Dr. Roux made an important discovery demonstrating that axonal transport was altered in the absence of MeCP2 and that cysteamine treatment, known to restore intracellular dynamics and BDNF secretion, improved lifespan and motor deficits in Rett mouse. More recently, he developed a project of genetic therapy using a codon-optimized mouse version of *Mecp2* under the control of a short *Mecp2* promoter, and in this proposal he is investigating whether *BDNF* gene therapy will be effective to bypass the challenges of *MECP2* gene therapy.

As a principal investigator in the field of Rett syndrome (RTT), Dr. Roux has authored more than 45 international peer-reviewed publications and has been the recipient of many national and international grants, including 2 Europeans networks (Dischrom, Eurorett). Dr. Roux is part of the Human Neurogenetics team (Head, Laurent Villard, PhD) at Inserm UMR 1251, Marseille Medical School (Faculté de Médecine de La Timone).