



## Orphan Drug Designation to sirolimus in Sickle Cell Disease

**Milan (Italy), January 24<sup>th</sup> 2018** – Rare Partners Srl Impresa Sociale announces that the European Commission has granted Orphan Drug Designation (ODD) to sirolimus (Rapamycin) for the treatment of sickle cell disease.

The decision, taken by the Commission on 18 January 2018, follows the positive opinion released by the Committee for Orphan Medicinal Products (COMP) of the European Medicines Agency (EMA), recommending the designation of the medicinal product, sirolimus, as an orphan medicinal product for the indication: treatment of sickle cell disease.

Sirolimus is now listed as an orphan drug for sickle cell disease in the Community Register of orphan medicinal products for human use with the code EU/3/17/1970. This designation follows previous designations by EMA and FDA regarding the use of sirolimus in beta-thalassemia patients.

Marco Prosdocimi, Managing Director of Rare Partners, said that *“We started the collaboration with professor Gambari and his group at the University of Ferrara focusing the attention on beta-thalassemia, realising soon that the results obtained by these outstanding scientists could be extended to another pathological condition, namely sickle cell disease. We therefore applied to EMA for an ODD in this new indication and COMP rapidly released a positive opinion, confirming that the clinical application of our experimental findings is fully logical and potentially useful for the patients. This important result further confirms that the collaboration we established since 2011 with Roberto Gambari and his research group is extremely fruitful”*.



EMA grants orphan medicinal product designations to advance the development of drugs intended to treat, prevent or diagnose life-threatening or very serious conditions that are rare and affect not more than 5 in 10,000 persons in the EU.

*“With this designation”, added Dr. Prosdocimi, “Rare Partners will have market exclusivity in the EU for ten years in the event that sirolimus receives marketing approval as a treatment for sickle cell disease”.*

This designation also gives right to special benefits, including research support, eligibility for protocol assistance, and possible exemptions or reductions in some regulatory fees during development or at the time of application for market authorization.

Roberto Gambari, Director of the Department of Life Sciences and Biotechnology, Ferrara University, said that: *“The effects of sirolimus and other inducers of fetal haemoglobin (HbF) is particularly relevant in the case of SCD, considering the fact that HbF retain anti-sickling effects. Our laboratory is at present focusing on the analysis of the effects on SCD erythroid cells of several molecules found to be HbF inducers in  $\beta$ -thalassemia; in addition, double heterozygous  $\beta$ -thalassemia/SCD patients are of great interest for optimizing treatment protocols. With this in mind, patient stratification is being considered for determining possible links between genomic polymorphism and response to the inducers, including sirolimus. This would greatly help in the process of patient recruitment in case that pilot clinical trials will be performed. The possibility of performing clinical trials based of this ODD should be considered of great importance for future developments. With respect to this achievement, we would like to mention the participation of our group to the THALAMOSS Project (THALAssaemia MODular Stratification System for personalized therapy of beta-thalassemia; Health-2012-INNOVATION-1) and the continuous support of Fondazione Cassa di Risparmio di Padova and Rovigo, which was the major national sponsor of our research activity on hemoglobinopathies.”*



## About Sickle Cell Disease

The term sickle cell disease (SCD) describes a group of inherited red blood cell disorders. People with SCD have abnormal hemoglobin, called hemoglobin S (HSS) or sickle hemoglobin, in their red blood cells. The disease affects the red blood cells, causing episodes of sickling, which produce episodes of pain and other symptoms. In between episodes of sickling, people with SCD are normally well. Long-term complications can occur. Certain conditions, such as infection, lack of fluid in the body (dehydration) or low oxygen, can trigger sickling.

## About sirolimus application in Sickle Cell Disease

It is known that an elevated level of fetal haemoglobin in SCD patients may result in a relevant clinical improvement. Sirolimus, already used as an immunosuppressant in transplanted patients, should act in SCD patients by inducing erythroid differentiation and expression of fetal haemoglobin, thus reducing the need of frequent blood transfusions. This new use of the drug has been patented by professor Gambari and co-workers. Rare Partners finalized an agreement with the assignees (University of Ferrara and Associazione Veneta per la Lotta alla Talassemia), aimed at completing the preclinical studies and proceeding with Orphan Drug Designation and clinical development in patients with haemoglobin pathologies.

## About University of Ferrara

The University of Ferrara, established in 1391, is one of the oldest universities in Italy, counting with more than 18,000 students and with an outstanding track-record of excellence in scientific research, including life sciences. Professor Roberto Gambari is the founder and Director of the Laboratory for the development of genetic and pharmacogenomic therapy of thalassemia, ThalLab, at the University of Ferrara. At



present, Roberto Gambari is Director of the Department of Life Sciences and Biotechnology, Ferrara University, and was the coordinator of the EU FP7 Project THALAMOSS.

## About Rare Partners

Rare Partners is a non profit biopharmaceutical company devoted to the development of new therapies and diagnostics in the field of rare diseases. The company was founded in Milan on March 2010 and registered in Italy as “Impresa Sociale”. The basic idea of Rare Partners is to match non profit financial resources (public and private) with industrial drug development expertise, provided by the company’s organization together with a strong network of consultants.

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