



**For the promotion  
and recognition  
of excellence  
in orphan disease  
treatment research**

### The Review Committee

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Milan, 4<sup>th</sup> September 2019

## **The Ninth edition of the International Prize for Scientific Research Arrigo Recordati awarded Alberto Auricchio's research project**

Today the Ninth edition of the International Prize for Scientific Research Arrigo Recordati came to its conclusion with a €100,000 research grant awarded to Professor Alberto Auricchio, MD, Professor of Medical Genetics at the Department of Advanced Biomedicine, "Federico II" University in Naples, and Coordinator of the Molecular Therapy Program at Telethon Institute of Genetics and Medicine (TIGEM) in Pozzuoli (Naples), Italy.

## **The International Prize for Scientific Research Arrigo Recordati**

The Prize was established in 2000 in memory of the Italian pharmaceutical entrepreneur Arrigo Recordati to carry on his legacy and to inspire researchers to make important discoveries benefiting people worldwide.

In the last ten years Recordati made the rare diseases field a health priority and increased its commitment worldwide through its dedicated subsidiaries to research, develop and market a number of treatments for orphan diseases.

To reflect its commitment, Recordati decided that the 2019 Edition of the International Prize for Scientific Research Arrigo Recordati be dedicated **to the promotion and recognition of excellence in orphan disease treatment research.**

International projects within the area of rare/orphan disease treatments in all therapeutic areas (except for oncology, hematology and immunology) were eligible to apply as long as the disease concerned has a prevalence of not more than 1 in 2000.

The 2019 Award was given in recognition of ongoing research projects, for which preliminary results, such as proof-of-concept and proof-of-principle, have already been obtained.

The 2019 Award was open to researchers of all nationalities who are not in any way directly affiliated with pharmaceutical or medical device companies. **The winner project will receive a research grant of € 100,000.**

**The Review Committee** is an independent panel of internationally recognized experts who have provided leadership throughout their long careers in the field of rare diseases.

The selection of the winning project was based on the evaluation of the Review Committee according to the pre-specified Award criteria, quality of the research as well as the therapeutic impact of the results.

All members of the Review Committee assessed the initial proposals submitted by forty-eight researchers of eighteen different nationalities using a two-step selection procedure.

The Review Committee evaluated the full proposals of the five most outstanding applicants and today they are delighted to announce that **Professor Alberto Auricchio's proposal was chosen as the winner of the € 100,000 Arrigo Recordati Prize.**

*"I would like to underline the high quality of the five finalist projects, the difficulty we had to choose between such various subjects"* said Robert Desnick, just before announcing Alberto Auricchio's project as the winner of the International Prize for Scientific Research Arrigo Recordati 2019.

*"We are more and more committed to meeting the needs of patients suffering from rare diseases by supporting research in this area" - said Andrea Recordati, CEO of the Recordati Group announcing the winner. "On behalf of the Recordati group, Robert Desnick, Francesco Emma, Norio Sakai and myself, it is an honour and a privilege to award Alberto Auricchio's outstanding research project that could potentially reduce the impact of an extremely severe rare disease and provide needed therapies".*

Alberto Auricchio, after expressing his satisfaction for the important Award, gave a rapid lecture summarizing his research project on **Gene therapy of Stargardt disease with AAV intein vectors.**

## Stargardt disease (STGD1)

**Stargardt disease (STGD1)** is a genetic (hereditary) disorder of the eye that leads to progressive loss of sight. The signs and symptoms of Stargardt disease typically appear in late childhood to early adulthood and worsen over time. Stargardt disease is caused by abnormalities in a gene called ABCA4. The ABCA4 gene is responsible for the production of a protein called ABCR that regulates the transport of substances in and out of some cells in the retina (the light-sensitive surface at the back of the eye).

In patients with Stargardt disease, ABCR does not work properly. This causes deposits to build up inside the retina cells, which become damaged and eventually die. Stargardt disease is a long-term debilitating disease because the patient's sight becomes progressively worse and eventually leads to blindness. Worldwide prevalence of STGD1 is estimated at 1/8,000 - 1/10,000.

## Gene therapy of Stargardt disease with AAV intein vectors

Coordinator of the Molecular Therapy Program at Telethon Institute of Genetics and Medicine Molecular, Professor Auricchio explains his research project: *"Our group has a long-standing interest in the development of gene therapies for inherited ocular diseases. Our research spans from tailoring the adeno-associated viral (AAV) vector platform to retinal gene transfer to proof-of-concept in animal models of retinal disease up to first-in-human. Indeed, we have importantly contributed to the phase I/II clinical trial of Luxturna, which is the first ocular gene therapy product on the market.*

*One of the limitations of AAV is its DNA cargo capacity of about 5 kb in size. This would not be sufficient for gene therapy of conditions like Stargardt disease (STGD1), the most common inherited macular degeneration in humans, which is caused by mutations in ABCA4, a gene with a coding sequence significantly larger than 5 kb.*

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*To overcome this, we have recently developed a system based on two AAV vectors, each encoding for one of the two halves of ABCA4 each flanked by short split-inteins which mediate protein trans-splicing and full length ABCA4 reconstitution in the retina of mice, pigs and in human retinal organoids. This system reduces lipofuscin accumulation in a mouse model of STGD1 and supports further development of AAV intein for therapy of STGD1 and other Inherited Retinal Diseases (IRDs) due to mutations in large genes.*

*The overall objective of the project funded by the International Prize for Scientific Research Arrigo Recordati is to translate this proof-of-concept of pre-clinical efficacy of AAV split intein for STGD1 into a first-in-humans by defining both AAV intein dose-response and safety in view of a future clinical trial. This will importantly contribute to the development of gene therapy for the common and severe STGD1” concluded Professor Auricchio.*

## 2019 Award Winner: Alberto Auricchio

Alberto Auricchio, MD is Professor of Medical Genetics at the Department of Advanced Biomedicine, “Federico II” University in Naples, and Coordinator of the Molecular Therapy Program at Telethon Institute of Genetics and Medicine (TIGEM) in Pozzuoli (Naples), Italy.

**His research is focused on gene therapy of retinal and metabolic diseases using adeno-associated viral vectors.** His group has contributed to the phase I/II clinical trial of Luxturna, the first approved gene therapy drug for an ocular disease, and to the development of gene therapy for mucopolysaccharidosis VI up to an ongoing phase I/II trial.

Professor Auricchio is co-author of more than 130 peer-reviewed publications on international scientific journals and inventor of several international patents on the use of viral vectors for gene therapy. He is a member of the editorial boards of various journals and of the European Society of Cell and Gene Therapy. He is a member of the editorial boards of *Molecular Therapy*, *EMBO Molecular Medicine* and *Translational Vision Science & Technology*.

In 2006 Alberto Auricchio received the Outstanding New Investigator Award from the American Society of Gene Therapy and in 2007 was nominated “Cavaliere of the Italian Republic” by the President of the Italian Republic. In 2011 he received the Consolidator Grant from the European Research Council - ERC - and the Advanced Grant in 2016.

## The Telethon Institute of Genetics and Medicine

The Telethon Institute of Genetics and Medicine (TIGEM), a Telethon Foundation organization, was founded in 1994 as a leading Italian research center. Located in Pozzuoli, Italy, just a few kilometers away from Naples, its new facility hosts over 200 staff members, including 18 research groups dedicated to understanding the molecular mechanisms behind rare genetic diseases.

These diseases, often overlooked by pharmaceutical industries, are most common in children and adolescents. TIGEM’s sole purpose is to provide the scientific basis for the development of treatments.

The Institute boasts three research programs, Cell Biology, Molecular Therapy and Systems Biology, 10 core facilities and significant international support in the form of funding and collaboration opportunities. TIGEM’s success is widely-recognized, resulting in a network of international support in the form of funding. Some of the major funding agencies include the European Union and its European Research Council (ERC). The ERC is currently funding five of the Institute’s faculty members with starter and advanced grants. Two of their projects are led by Professor Alberto Auricchio.

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## Recordati and Rare Diseases: focused on the few

Recordati is an international pharmaceutical group dedicated to the research, development, manufacturing and marketing of pharmaceuticals. Headquartered in Milan, the Group has a staff of more than 4,100. Recordati has grown constantly since 1926 thanks to the quality of its products and to the implementation of a decisive internationalization policy through a focused strategy of acquisitions and license agreements. Recordati produces and promotes innovative medicines worldwide including both products for specialty and primary care therapies and treatments for rare diseases.

The Group operates in the rare disease segment worldwide through its dedicated subsidiaries in Europe, in the Middle East, in the U.S.A., Canada, Russia, Australia, Japan and in some Latin American countries. Recordati constantly enhances its therapeutic offering through the development of its product pipeline with a focus on rare diseases, mainly treatments for metabolic deficiencies of a genetic nature.

Recordati is committed to supporting families affected by these diseases through the research and development of new therapies and through the diffusion of specific knowledge within the medical community.

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