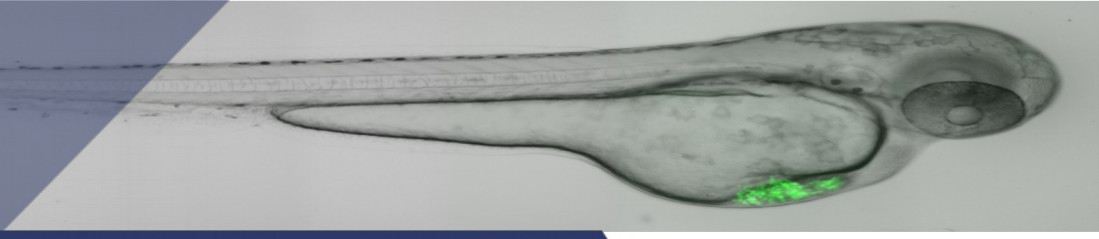
TECH PROFILE



Transgenic non-human models

Business area	Market sector	Medical Indication
Discovery	*Obesity*	*Obesity, Morbid obesity*

FUNDACIÓN INSTITUTO DE INVESTIGACIÓN SANITARIA SANTIAGO DE COMPOSTELA

Research goal

Lipodystrophies & PELD treatment

Problem to solve

Lipodystrophies are characterized by the lack of fat, which is associated with insulin resistance, diabetes, dyslipemia and fatty liver. One of the most-severe form of lipodystrophy, type 2 congenital generalized lipodystrophy (CGL2), is caused by variants in BSCL2/seipin. Seipin is a protein encoded by the BSCL2 gene. Other diseases are associated with BSCL2, as PELD (Progressive Encephalopathy with or without Lipodystrophy), a subtype of CGL2, a pediatric fatal disease characterized by lipodystrophy and severe neurodegeneration. These children die before 10 as a consequence of neurodegeneration and uncontrolled seizures. At present there is not cure for PELD nor for CGL2. The present invention is focused on solving the above mentioned problems and, consequently, it is herein provided transgenic non-human models for lipodystrophy and PELD which can be used for screening candidate compounds to be used as drugs for treating insulin resistance, fatty liver, lipodystrophy and PELD.

Innovation

Our mice incorporate a transgene that does not exist as such in healthy humans, and make them unique in the market, showing two qualities: their versatility and the possibility of serving as models for the study of common diseases such as diabetes and fatty liver, as well as rare diseases such as lipodystrophy and PELD. Specifically, our C57BL/6N Bscl2$^{-/-}$ mouse in homozygosity presents lipodystrophy, impaired glucose metabolism and fatty liver, but if crossed with Cre mice available on the market, the global or tissue specific mouse C57BL/6N BSCL2$^{Celia/Celia}$ can be obtained. There are hardly murine models in the market for lipodystrophy and none for PELD, an interesting model of neurodegenerative disease. Our mice are excellent models for studying common diseases (insulin resistance, diabetes, fatty liver), but also rare disorders as lipodystrophy and PELD.

Market opportunity

Academia: Many groups are devoted to unravel the molecular bases of insulin resistance, fatty liver, neurodegeneration and testing drugs in pre-clinical trials, and more and more groups are working in lipodystrophy, which would be interested in having our mice. Pharma: Our mice are unique for the testing pre-clinical therapeutic approaches. Repositories of transgenic mice: they provide mice for studying different diseases and for pre-clinical trials both from Academy and from pharma industry.

Research team

Molecular Endocrinology Research Group of the Health Research Institute of Santiago de Compostela:

- **David Araújo-Vilar & Sofía Sánchez-Iglesias**: PIs of the project.
- **David Araújo-Vilar** : Senior consultant endocrinologist at University Clinical Hospital of Santiago de Compostela.

Intellectual property

EP19382828.2 "TRANSGENIC NON-HUMAN MODELS FOR LIPODYSTROPHY AND/OR PELD"

Development stage:

Available for: *Licensing, co-development*

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