GOBIERNO DE ESPAÑA
MINISTERIO DE HACIENDA
SECRETARÍA DE ESTADO DE PRESUPUESTOS Y GASTOS
DIRECCIÓN GENERAL DE FONDOS EUROPEOS

Generalitat de Catalunya

UNIÓN EUROPEA

Una manera de hacer Europa

BUENAS PRÁCTICAS
Actuaciones Cofinanciadas

"GenomicScientia, Centre for Genomic Sciences in Medicine".
Sant Joan de Déu Foundation.

Programa Operativo de Cataluña

Año 2018

Fondo Europeo de Desarrollo Regional

We present as Good Practice the project "GenomicScientia, Centre for Genomic Sciences in Medicine" by the Sant Joan de Déu Foundation.

The project "GenomicScientia, Centre for Genomic Sciences in Medicine" aims to learn about the genetic causes of rare diseases and patients affected by undiagnosed diseases. Therefore, with ERDF's joint funding, a special structure has been built to carry out translational research in Sant Joan de Déu Foundation. The objective is to transfer the knowledge obtained through basic research for preventing and treating problems detected at the clinic. In particular, the centre scientifically and technologically supports patients affected by a rare disease, especially children affected by a undiagnosed disease.

The Centre for Genomic Sciences in Medicine supports scientific and clinical activities carried out by the Foundation and Sant Joan de Déu Hospital, Universitat de Barcelona's children's hospital. Being fully committed to rare diseases, the Hospital created the Rare Diseases Paediatric Institute, a cross programme that was designed to comprehensively treat these diseases, that can also be applied in other health or academic institutions, thanks to the creation of the Centre for Genomic Sciences in Medicine, with special focus on undiagnosed diseases.

The launch of GenomicScientia represents an unprecedented knowledge hub for Catalan, Spanish and European paediatric medicine, converting Sant Joan de Déu Hospital into a highly competitive and unique centre that will benefit not only its own institutions, but will also serve as a reference centre for genomic medicine and rare diseases. GenomicScientia will also serve as a hub to attract the pharmaceutical and biotechnological industries, which will add great economic value.

This project had a total eligible cost of 1,024,370.00 €, with an ERDF support of 512,185.00 €. Fourteen researchers took part in the project (11 women, 3 of which were newly hired, and 3 men).

This operation is presented as Good Practice because it meets the following criteria:

1. **High level of dissemination among beneficiaries, potential beneficiaries and the general public**

The project GenomicScientia has been properly disseminated among beneficiaries, potential beneficiaries and the general public, relying on Sant Joan de Déu Foundation's department of communication, which engages in giving visibility to biomedical and social research related to Sant Joan de Déu's centres.

Below are some examples of activities that were carried out:

- **Sant Joan de Déu Foundation website**

Announcement on the website concerning the awarding of the project and explaining its objectives and overview of the awarded project on the Neurogenetics and Molecular Medicine group's project section:

ERDF's co-funding is highlighted with the sentence "This project has been co-funded by the European Union through the European Regional Development Funds (ERDF)".

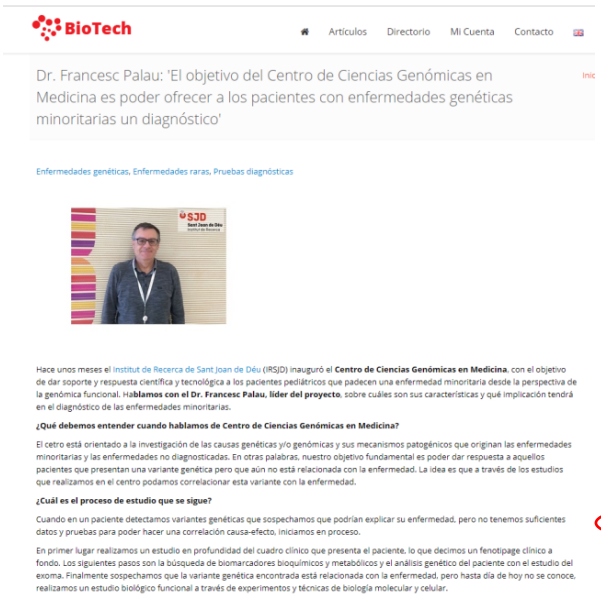
Furthermore, the interview with Chief Investigator Dr. Francesc Palau is also notable: "The objective of the Centre for Genomic Sciences in Medicine is to offer diagnosis to patients affected by rare genetic diseases". The interview was published on the Foundation's website and it was also shared through social media.

Twitter: Tweet reporting on the Centre for Genomic Sciences in Medicine with the hashtag #FEDERrecerca, also mentioning @gencat.

Facebook: Publication reporting on the Centre for Genomic Sciences

➢ External actions

The interview was also published on the BioTech website:



BioTech

Artículos Directorio Mi Cuenta Contacto

Dr. Francesc Palau: 'El objetivo del Centro de Ciencias Genómicas en Medicina es poder ofrecer a los pacientes con enfermedades genéticas minoritarias un diagnóstico'

Enfermedades genéticas, Enfermedades raras, Pruebas diagnósticas

Hace unos meses el Institut de Recerca de Sant Joan de Déu (IRSJD) inauguró el **Centro de Ciencias Genómicas en Medicina**, con el objetivo de dar soporte y respuesta científica y tecnológica a los pacientes pediátricos que padecen una enfermedad minoritaria desde la perspectiva de la genómica funcional. Hablamos con el Dr. Francesc Palau, líder del proyecto, sobre cuáles son sus características y qué implicación tendrá en el diagnóstico de las enfermedades minoritarias.

¿Qué debemos entender cuando hablamos de Centro de Ciencias Genómicas en Medicina?

El cetro está orientado a la investigación de las causas genéticas y/o genómicas y sus mecanismos patogénicos que originan las enfermedades minoritarias y las enfermedades no diagnosticadas. En otras palabras, nuestro objetivo fundamental es poder dar respuesta a aquellos pacientes que presentan una variante genética pero que aún no está relacionada con la enfermedad. La idea es que a través de los estudios que realizamos en el centro podamos correlacionar esta variante con la enfermedad.

¿Cuál es el proceso de estudio que se sigue?

Cuando en un paciente detectamos variantes genéticas que sospechamos que podrían explicar su enfermedad, pero no tenemos suficientes datos y pruebas para poder hacer una correlación causa-efecto, iniciamos en proceso.

En primer lugar realizamos un estudio en profundidad del cuadro clínico que presenta el paciente, lo que decimos un fenotipage clínico a fondo. Los siguientes pasos son la búsqueda de biomarcadores bioquímicos y metabólicos y el análisis genético del paciente con el estudio del exoma. Finalmente sospechamos que la variante genética encontrada está relacionada con la enfermedad, pero hasta día de hoy no se conoce, realizamos un estudio biológico funcional a través de experimentos y técnicas de biología molecular y celular.

demostrar la causa asociada a un gen determinado y lograr el diagnóstico genético definitivo.

En que consiste este último paso en el estudio, ¿qué sería la genómica funcional?

Cuando en el paciente detectamos variantes genéticas que creemos podrían explicar la enfermedad, pero que aún no se ha estudiado en profundidad y desconocemos si tienen relación, requerimos de una validación analítica mediante una serie de estudios. Con estos estudios podemos analizar los efectos biológicos de las variantes y/o mutaciones en sistemas experimentales celulares y moleculares, con la finalidad de demostrar si aquella variante es o no es la causa de la enfermedad.

¿Finalmente podríamos decir que tenemos una descripción completa de la enfermedad?

Pues sí, toda la información que obtenemos a través de las aproximaciones y técnicas mencionadas nos da una visión global de la fisiopatología de la enfermedad, desde el análisis del genoma, la caracterización biológica funcional de las variantes genéticas encontradas, hasta el fenotipo y la expresión clínica de los pacientes.

Y además para poderlo hacer posible, contamos con la implicación de diversos profesionales. Desde el equipo clínico que realiza el primer contacto con el paciente, el personal de los laboratorios de bioquímica y genética clínica; y finalmente nuestro grupo de investigación, coordinado por la Dra. Janet Hoenicka en el IRSJD, que realiza toda la parte de genómica funcional.

¿Qué significa que el centro es una instalación singular?

Hablamos de instalación singular porque nuestro objetivo es sistematizar este proceso y poderlo ofrecer dentro de la cartera de servicios del Hospital Sant Joan de Déu. La idea es acoplarlo dentro de la dinámica del hospital, especialmente en el proceso de diagnóstico, haciendo realidad el concepto de investigación traslacional en medicina.

Imaginémonos un paciente que llega al hospital con una determinada enfermedad y el equipo clínico sospecha que puede ser genética. En primer lugar le haríamos un análisis genético, si en esta prueba vemos que presenta una variante no descrita pero que sospechamos que puede explicar la enfermedad, pasaríamos a hacer todo el análisis experimental para encontrar la respuesta.

En estos momentos me atrevería a decir que el Hospital Sant Joan de Déu es pionero en el país en ofrecer este tipo de servicio traslacional.

¿Qué impacto tendrá el Centro de Ciencias Genómicas en Medicina en los pacientes?

El principal beneficio para los pacientes es que conseguiremos resolver su diagnóstico, es decir le podremos decir al paciente y si familia cuál es la mutación que origina la enfermedad cuando es evidente. De esta forma, podremos ofrecer a las familias asesoramiento genético, a través del riesgo de recurrencia de la mutación en la familia. Y en último lugar aplicar determinados procesos terapéuticos en función de la mutación que presenten y su expresión fenotípica.

Pero debemos pensar que para los paciente y las familias es muy importante poder poner nombre a su enfermedad y tener un diagnóstico, aunque no haya un tratamiento disponible.

Este proyecto ha sido financiado por la Unión Europea a través del Fondo Europeo de Desarrollo Regional FEDER (Una manera de hacer Europa) con el soporte de la Generalitat de Cataluña.

Nº de referencia: 2015 FEDER-SGT

Ayuda concedida: 512.185€

And a tweet was published on Sant Joan de Déu Laboratory's profile:

Laboratori SJD Laboratori
@SJDLaboratori

Siguiendo

'El objetivo del Centro de Ciencias Genómicas en Medicina es poder ofrecer a los pacientes con enfermedades genéticas minoritarias un diagnóstico'

"El objetivo del Centro de Ciencias Genómicas en Medicina...
Hace unos meses el Institut de Recerca de Sant Joan de Déu (IRSJD) inauguró el Centro de Ciencias Genómicas en Medicina, cofinanciado con Fondos FEDER y de la Generalitat de
fsjd.org

9:33 - 23 may. 2018

1 Retweet 3 Me gusta

➢ Signs

Sign at the entrance of the Foundation's laboratory

Sign at the entrance of the hospital's laboratory

2. Inclusion of innovative elements

With this genomic diagnosis and research platform, cell and molecular biology is incorporated into the clinical practice of diagnosing rare diseases as an innovation. It can be summarised in two specific actions for genetic diagnosis: analysing the cell expression and functions of genetic variations of candidate genes as part of the diagnosis process in undiagnosed children; and integrating biomedical research in the clinical practice as part of a translational model that fosters interaction between clinic doctors, laboratory clinic geneticists and researchers, all within the Rare Diseases Paediatric Institute, in order to give a scientific solution to treatment needs.

3. Linkage between results obtained from the supported actions and the objectives established

The project has changed the scientific diagnosis process of rare diseases, especially neurological diseases, which are the most significant group. The project has provided a top-rated technological and scientific tool to complement the comprehensive clinical approach towards assistance to patients affected by rare diseases and their families, all of this as part of Sant Joan de Déu Hospital. The project has allowed for developing an optimal translational research model, by bringing cell and molecular biology closer to scientific issues arising from clinical practice.

4. Contributing to resolving a regional problem or weakness

Rare diseases genetic diagnosis is a problem affecting the whole of Catalonia. Sant Joan de Déu Hospital, as a reference paediatric centre for the treatment of complex pathologies, has made an effort with the Centre for Genomic Sciences in Medicine to improve and accelerate genetic diagnosis in patients, reducing the time this long journey takes until a diagnosis is made.

In particular, the model 'clinical genomics – functional genomics' developed as part of this project, allows for offering genetic diagnosis and genetic counselling to patients, for whom genetic analysis provided information that is not definitive. This translational model, which lies somewhere between clinical practice and biomedical research, is applied to selected patients from a total of 17,000 rare diseases patients of the Sant Joan de Déu Hospital, the majority of whom are Catalan. It represents a qualitative leap that provides a solution to and guides those patients and the families of patients who were not (or are not) diagnosed, with the corresponding immediate consequences to their lives, within the family and on a social level.

5. High degree of coverage of the target population

This project impacts Catalonia's paediatric population that is taken care of at Sant Joan de Déu Hospital (more than 50% of this population). Additionally, it has a direct effect on families (mostly parents) with regard to their own genetic diagnosis, but especially to the genetic counselling, as well as the possibility to offer couples with children affected by genetic rare diseases the option of prenatal diagnosis or preimplantation genetic diagnosis.

6. Compliance with horizontal principles (sustainable development, equality between men and women and the principle of non-discrimination) and environmental legislation

Sant Joan de Déu Foundation has been acknowledged by the Human Resources Strategy for Researcher, an emblem awarded to institutions that apply the European Charter for Researchers and a Code of Conduct for the Recruitment of Researchers. This emblem aims at contributing to developing an attractive European job market by granting equal opportunities to researchers (women/men) and encouraging the incorporation of women into the research sector. We also have a Prevention and Action Protocol in case of labour harassment and discrimination, which includes objectives, scope of application and an action process in the case a report is filed.

With regard to the environment, we strive to avoid that the activities carried out by the Foundation negatively affect the natural environment, continuously improving our environmental behaviour. For this reason, we have a Waste Plan, essentially based on reducing the potential risk to people's health and the environment, stemming from a deficient waste identification, management, transport and storage; minimising costs of global waste management; taking appropriate protection measures to manage hazardous waste and provide laboratory employees with comprehensive information to correctly classify waste, by placing waste in the corresponding trash bin or container.

Lastly, in accordance with current legislation of non-discrimination, the Sant Joan de Déu Foundation currently employees three people with disabilities and as an additional measure, it works with a special work centre.

7. Synergies with other policies or instruments of public intervention

The Centre for Genomic Sciences in Medicine (GenomicScientia) made it possible for the research group to develop new projects that have received regional and national funding from both the public and the private sectors. The research group has been able to secure resources for projects, and also to incorporate human resources in national public funding tenders with the Health Institute Carlos III or the Ministry of Science and Innovation, as well as private tenders, such as the Foundation Ramón Areces or the Foundation Isabel Gemio. They also accessed regional funding tenders, and lastly, they have received non-competitive funding, predominantly from associations of patients, such as the Foundation Amigos de Nono.

On the other hand, GenomicScientia promotes more added value activities in the field of rare diseases and undiagnosed diseases, which are a scientific and social challenge. The project has helped establish a scientific basis within hospital fields at the service of patients which allows for the scientific approach to the resolution of clinical issues. The project results focus on people's wellbeing in the health sphere, and it implies a direct benefit for and improvement in the quality of life of the people affected by these diseases and their families, and as a result also for society. This fact perfectly lines up with the third objective of the RIS3CAT strategy (the research and innovation strategy for the intelligent specialization of Catalonia), favouring the creation of knowledge in the field of rare diseases and undiagnosed diseases, and also strengthens Sant Joan de Déu Foundation's, and also Catalonia's, positioning nationwide and Europe-wide, boosting the health sector, which has been mentioned as one of the key sectors in Catalonia.

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