Rare Disease Day

29/02/2024

On this Rare Disease Day, 29th of February, we would like to encourage you to watch this video prepared by the European Joint Programme on Rare Diseases from the European Union (EJP RD), in

(Click on the image, Dr. Vicente Andrés, as he explains the ProgerOmics project at 7:08).

which the CNIC participated as the coordinator of the ProgerOmics project led by Dr. Vicente Andrés. EJP RD brings together over 130 institutions (including all 24 ERNs) from 35 countries to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care, and medical innovation. EJP RD has been instrumental in addressing this challenge by providing funding and support for ProgerOmics.

Hutchinson-Gilford progeria syndrome (HGPS) is an extremely rare genetic disorder, affecting one person in 18 million. It is characterized by accelerated aging and premature death with no current cure. Diagnosis typically occurs at 1-2 years of age, and patients succumb to complications of atherosclerosis, such as heart failure or stroke, around the age of 14.5 years. The progression of HGPS varies widely among individuals, making clinical assessment the primary tool for monitoring disease advancement and treatment response.

This project will help to address this gap by employing a comprehensive biomarker discovery approach, utilizing multi-omics studies in progeroid mice to identify robust biomarkers of HGPS progression. The project involves expert partners and three HGPS patient associations, emphasizing patient engagement and dissemination of results. The ultimate goal is to enhance personalized medicine for HGPS patients through improved monitoring using circulating biomarkers. The "FAIR" Data Management Plan ensures open access to generated data, facilitating clinical translation.

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