SUCCESS STORIES


Chkniti Khongchum

DIAGNOSIS AND EVOLUTION OF RARE DISEASES

The exact number of rare diseases in existence is unknown, but it is somewhere between 5,000 and 7,000. Due to their infrequency, these rare diseases can take years to be diagnosed, in what is known as a diagnostic odyssey, involving an endless series of hospital tests. Here, genomics can help, speeding up the diagnostic process. Through genome analysis, new genomic data can be generated to diagnose patients and predict how a disease will develop. This is what is being done by the Bioinformatics Unit and Data Analysis Team, one of the research branches of the National Genomic Analysis Centre (CNAG-CRG, www.cnag.crg.eu). By sequencing the DNA of patients with rare diseases, they develop plans to process data and tools, such as the RD-Connect GPAP platform. They then pass these on to doctors and geneticists, so that they can view and interpret the information in order to identify the defective part of the DNA that is causing the disease and find a diagnosis. “For this data to be understood, experts from different fields are required”, explains Leslie Matalonga, a clinical genomics specialist and member of the group. They participate in various initiatives (such as URDCat, Solve-RD and Matchmaker Exchange) involving various hospitals and research centres on a national and international level. These are platforms with data from patients and their family members. Each person’s data is analysed individually and information is linked thanks to algorithms that detect similarities, contribute towards diagnosing new patients, and help to put patients’ families around the world with the same disease in touch.

COMPOSITE MATERIALS IN RENEWABLE MARINE ENERGIES

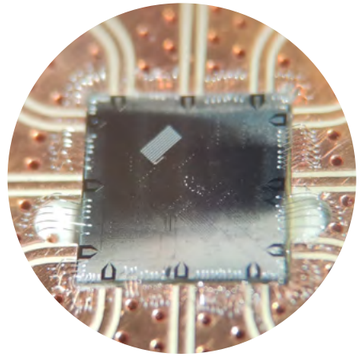
Until now, steel has been the most commonly used material for turbines on floating offshore wind platforms, despite being affected by corrosion due to the marine environment. This is one of the reasons why the International Centre for Numerical Methods in Engineering (CIMNE) has been looking for new, more resistant composite materials, which combine two or more materials to produce a more sophisticated result. Naval engineer and head of CIMNE’s (www.cimne.com) Naval Engineering Department, Borja Serván Camas, is also the coordinator of Fibregy(fibregy.eu), a European project within the H2020 programme made up of more than 10 partners, including research centres, universities, private companies, shipyards, composite material manufacturers’ associations, and a ship classification society. The project’s goal is to encourage wider implementation of these materials for use on offshore renewable energy collection platforms. “We have various objectives, such as generating a digital twin to make structural calculations regarding behaviour at sea, or selecting composite materials based on resistance, structural integrity, recyclability, usability, and environmental and economic criteria”, Serván explains. There is an extensive catalogue of composite materials being used in very different disciplines, but here, tests are only being conducted with resins, fibreglass and carbon fibre. There are plans to build new platform structures with composite materials and install them on the surface to replace the existing ones, thus demonstrating that these new prototypes are viable in this kind of environment.

DISCOVERING THE ORIGIN OF PLANT DOMESTICATION

In the current context, with climate change and all the stress it is putting on ecosystems, the goal is to maximise crop yields with the minimum possible inputs (water, land and pesticides). Studying plant genomes from an evolutionary standpoint can reveal which genes make ancient plants resistant to drought, for example, so that this gene can be found in modern plants in order to create new, more sustainable, better adapted varieties. “The idea is to reintroduce this gene, which was present in varieties from the past, to modern varieties, through traditional cross breeding programmes or, if possible, with CRISPR technology”, explains Laura Botigué, a Ramon y Cajal researcher at the Centre for Research in Agricultural Genomics (CRAG, crag.cat), whose work involves carrying out genomic analyses of ancient plant samples. She works with archaeobotanists and museums, who provide her with archaeological plant samples. She then extracts DNA from them in order to sequence it and analyse the plants’ genome. This research has a historical aspect that sheds light on the plant domestication process: “When I analyse the genome and compare it with the wild variant of the plant, I try to deduce whether there was one or more centres of domestication and hybridisation processes, or whether various varieties were selected in different places”, says Botigué. There is also a more agronomic side to the work, as it helps scientists to understand the process of plant dispersion and how important species like wheat, chickpeas, peas, lentils and beans were domesticated: “As these species spread across new territories, they adapted perfectly to new ecosystems”.

THE REASONS BEHIND MOBILITY IN ADULTHOOD

In a globalised world like our own, mobility experiences are diverse and complex, and a series of social and economic inequalities emerge in residential and geographical mobility patterns. “Until now, mobility research has focused on childhood or adulthood, but it has never sought a connection between the two”, indicates Sergi Vidal, a researcher at the Demographic Studies Centre (ced.cat) who will begin the project *Understanding spatial mobility from early life into adulthood* with the ERC Consolidator Grant in November. The aim of his research for the next five years is to have a better idea of why people move about, to understand the diverse nature of mobility patterns from childhood to adulthood, and to analyse their impact. “We know that mobility in childhood has an impact on children’s development, on their social relationships and on their educational performance, but we have not worked out the extent to which it can have consequences in adult life, or whether it can have an effect on our inclination to move around, on our career, on family relationships or on health”. Depending on the questions answered, certain databases will be used and a methodology will be established to utilise the data and take evidence from it: “We will develop machine learning methods to get a better understanding of this data, to see the mobility patterns that have been followed, and to examine how they are related to certain consequences in adult life”, Dr Vidal concludes.



THE FIRST QUANTUM COMPUTER PROTOTYPE IN SOUTHERN EUROPE

Since the quantum laws of nature were discovered in the 1920s, scientists have been looking at ways in which quantum physics could have practical applications. Now, in 2022, we know that a controllable quantum system can conduct simulations of real quantum systems in order to understand natural phenomena better or investigate how new medicines, fertilisers and other products could be designed. Quantum computers will be able to carry out mathematical calculations more efficiently than conventional computers, thus leading to optimal solutions and calculations that we cannot yet imagine. The first countries with quantum computers are likely to be the United States and China, and “they will not want to outsource them, because they will constitute a significant technological advantage over the rest of the world”, predicts Pol Forn-Díaz, a quantum physicist, head of the Quantum Computing Technology group at the Institute of High Energy Physics (IFAE, according to its initials in Catalan), and co-founder of the company Qilimanjaro Quantum Tech (www.qilimanjaro.tech), which aspires to commercialise quantum computing. The aim of this spin-off is to build a quantum computer and make it available to everyone, offering the service on both an academic and an industrial level, with an advisory team helping to tackle problems using quantum computing. In the future, quantum processors with millions of qubits will be able to implement sophisticated quantum algorithms. For now, Qilimanjaro Quantum Tech is a pioneer in southern Europe, having built a simple prototype with two quantum bits, with an experiment with five qubits in the pipeline.