On the lookout for the genes behind disease

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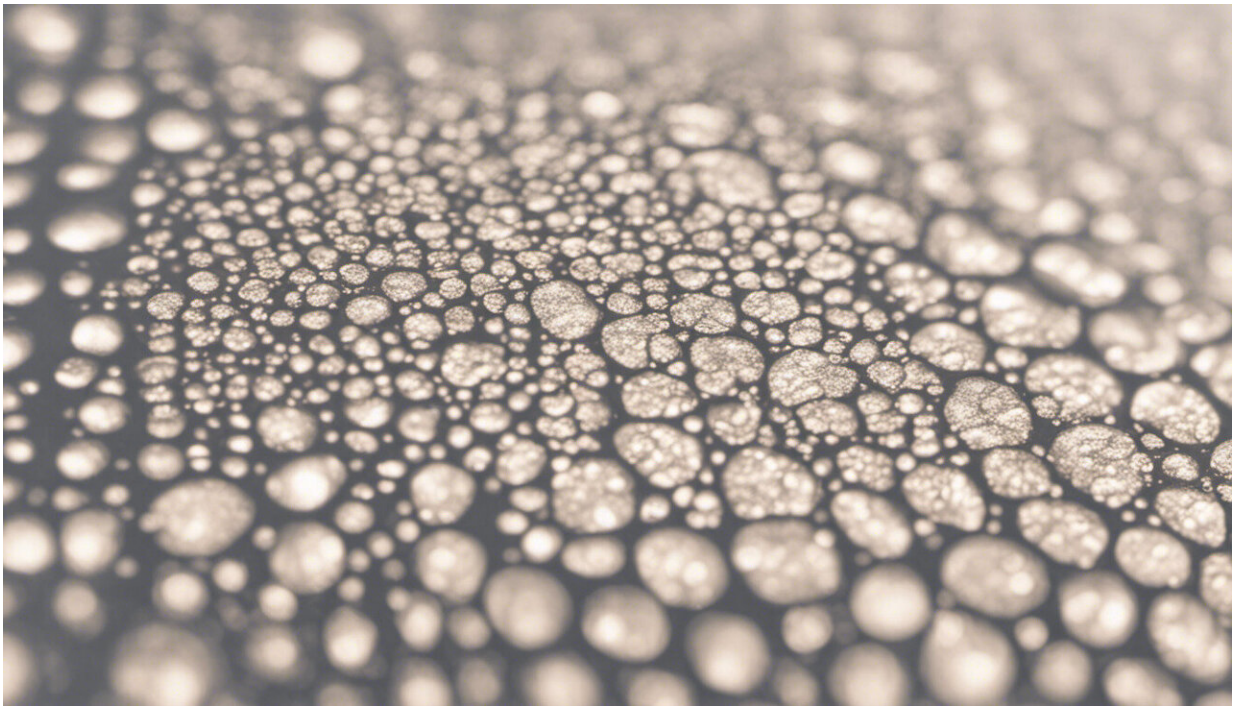

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Today's information age has provided the tools for biology to yield huge numbers of DNA sequences from many different species. Modern technology has made DNA sequencing simpler, less expensive and more reliable - with huge benefits for diagnosing and treating medical problems.

Previously the challenge was to collect genetic data. Today the challenge

lies in making sense of it.

"We're taking an evolutionary approach to making sense of DNA sequences - that means we examine the way that genes have evolved in order to understand better how they work," says Professor Aoife McLysaght of Trinity College Dublin, Ireland. She received a European Research Council (ERC) Starting grant worth around EUR 1.4 million for the project.

With her EU project DOSE ('Dosage sensitive genes in evolution and disease'), McLysaght is investigating differences between gene doses - that is, having more or less of the gene and the resulting effect on health.

"Changes in the amount of a gene between individuals - dose changes - are a relatively recent discovery and are sometimes implicated in disease," explains McLysaght. In simple terms, she has taken an evolutionary approach to figuring out which dose changes are acceptable, and which are likely to be involved in human disease.

For McLysaght this makes sense if you think about evolution as a large-scale natural experiment. She explains how over evolutionary time, just about every DNA change and recombination has been tried out, but only those that allowed us to survive were retained.

"By looking at evolution we can understand the acceptable and unacceptable changes to DNA," says McLysaght. "DNA changes that were unacceptable during evolution are likely to be the same as the ones that cause disease today."

By recruiting a team of talented post-doctorate and PhD students, McLysaght is hoping to bring together many different kinds of evolutionary and genomic information in order to make a sophisticated prediction of the dosage effects of any gene in the human genome.

"In this way, we will accelerate the discovery of disease genes," she says.

Although the project is still in its early stages and not slated for completion before the end of 2017, McLysaght and her colleagues have already published a paper on their research. The publication, 'Genome-wide deserts for copy number variation in vertebrates', outlines their strategy for homing in on diseased genes.

The team explains how DNA alterations of a genome, known as copy-number variations (CNVs) - which represent duplicated or deleted genes - are frequently associated with human disease. "We are hopeful that we will make a really significant contribution during the five years of this project," says the professor.

By enriching the information used in pinpointing disease genes, the team hopes to accelerate the detection process and make it less expensive. This has huge potential for more accurate diagnostics, which represents the first step towards better therapies. The new genetic strategy could pave the way to a whole new paradigm in treating congenital diseases - from blindness to osteoarthritis.

More information: Project factsheet
cordis.europa.eu/projects/rcn/104726_en.html

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