Scientists find link between genes and ability to exercise

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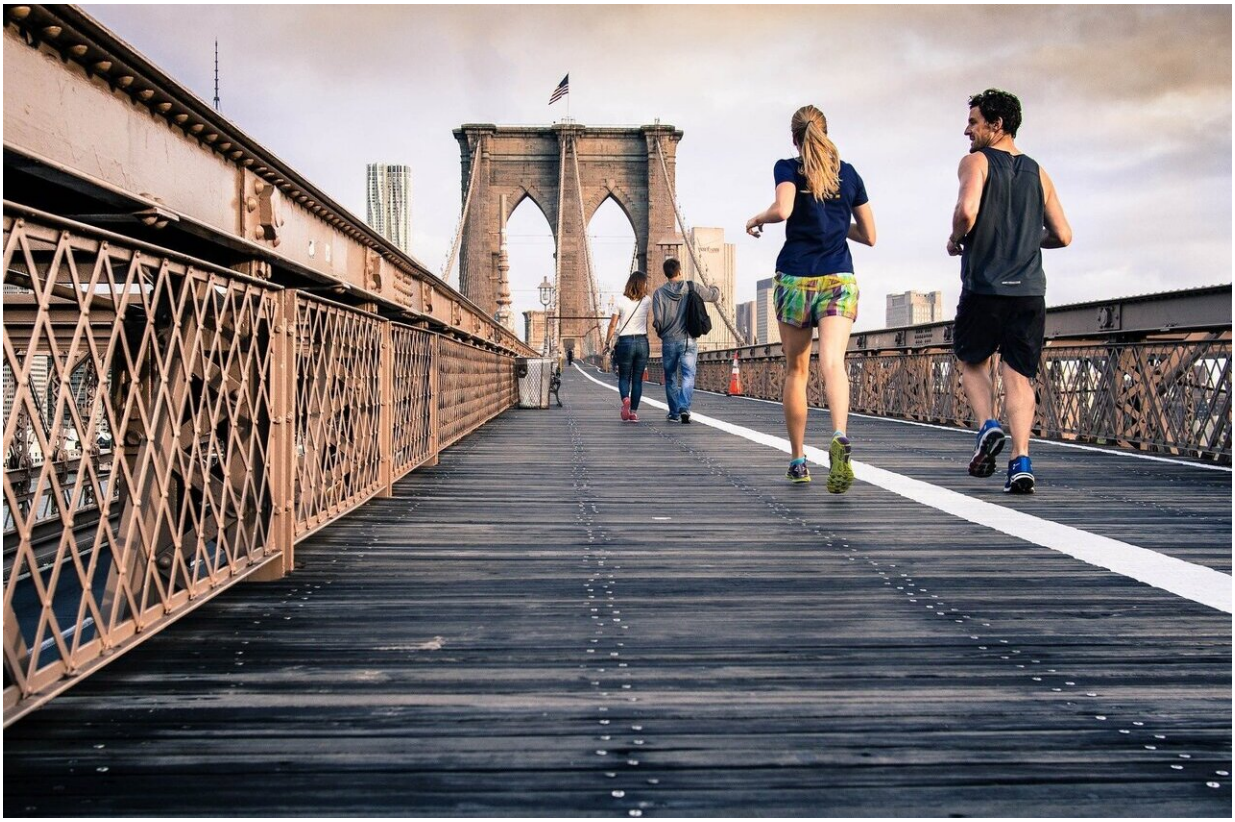

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A team of researchers have discovered a genetic mutation that reduces a patient's ability to exercise efficiently.

In a study published in The *New England Journal of Medicine*, a team including researchers from King's College London have found a link between a genetic mutation that affects cellular oxygen sensing and a patient's limited exercise capacity.

The team identified a patient who had a reduced rate of growth, persistent low blood sugar, a limited exercise capacity and a very high number of red blood cells.

The team carried out genetic and protein analysis of the patient, examined their respiratory physiology in simulated high altitude, measured their exercise capacity, and performed a series of metabolic tests.

The von Hippel-Lindau (VHL) gene is fundamental for cells to survive when oxygen availability is reduced. Following genetic analysis, an alteration on the VHL gene was identified and associated with impaired functionality in the patient's mitochondria, the powerhouse of the cell that uses oxygen to fuel cellular life. This reduced mitochondrial function efficiency limits the patient's aerobic exercise capacity compared to people without the mutation.

Dr. Federico Formenti, School of Basic & Medical Biosciences, one of the leading authors of the study, comments: "The discovery of this mutation and the associated phenotype is exciting because it enables a deeper understanding of human physiology, especially in terms of how the human body senses and responds to reduced oxygen availability."

A new syndrome has been discovered that can alter the regulation of human metabolism and skeletal muscle function. This research puts the basis for the study of new mutations that affect the oxygen sensing pathways and the way these mutations are associated with the integrative function of the human body as a whole. Improving our understanding of

these mechanisms may also contribute to the treatment of hypoxic conditions.

Provided by King's College London