Genetic Variants and Critical illnesses

The article is related to General Studies-Paper-III (Science & Technology)

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The genetic variants causing severe COVID-19.

The GenOMICC project is a research study that brings together clinicians and scientists from around the world to find the genetic factors that lead to critical illnesses.

To understand the genetic causes of severe COVID-19, the DNA of 7,491 critical patients was compared with 48,400 people who had not suffered from COVID-19, and that of a further 1,630 people who had only experienced mild symptoms.

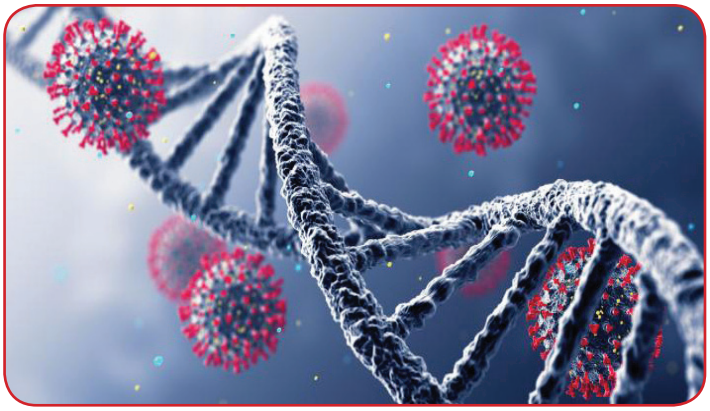
The study found key differences in 16 genes in ICU patients compared to the DNA of other groups. The new variants included some that had a role in blood clotting, immune response and the intensity of inflammation.

Jacob Koshy

The story so far: Scientists in the United Kingdom as part of a research project, GenOMICC (Genetics of Mortality in Critical Care), have identified 16 new genetic variants that make a person more susceptible to a severe COVID-19 infection.

What is the GenOMICC study?

The GenOMICC— reportedly the largest of its kind — is a research study that brings together clinicians and scientists from around the world to find the genetic factors that lead to critical illnesses. While millions suffer from infectious diseases every year, even though most cases are mild, some people become extremely unwell and need critical care. This may be because of their genes and the GenOMICC project is about identifying them. The scientists involved compare the DNA of critically-ill patients with members of the general population.



However, ferreting out such differences requires a large number of people and comparing their genetic structures at multiple levels of resolution. Since 2015, the GenOMICC has been studying emerging infections such as SARS (severe acute respiratory syndrome), MERS (Middle East respiratory syndrome), flu, sepsis, and other forms of critical illness.

How was the GenOMICC study for COVID-19 done?

Researchers from the GenOMICC consortium, led by the University of Edinburgh in partnership with Genomics England, sequenced the genomes of 7,491 patients from 224 intensive care units in the United Kingdom. Their DNA was compared with 48,400 other people who had not suffered from COVID-19, and that of a further 1,630 people who had experienced mild symptoms. Determining the whole genome sequence for all participants in the study allowed the team to create a precise map and identify genetic variation linked to severity of COVID-19.

What are the key findings?

The team found key differences in 16 genes in ICU patients compared to the DNA of the other groups. It also confirmed the involvement of seven other genetic variations already associated with severe COVID-19 discovered in earlier studies by the same team. The 16 new genetic variants included some that had a role in blood clotting, immune response and the intensity of inflammation. A single gene variant, the team found, disrupted a key messenger molecule in immune system signalling — called interferon alpha-10 — that increased a patient's risk of severe disease. There were variations in genes that control the levels of a central component of blood clotting — known as Factor 8 — that were linked with critical illness in COVID-19. This highlights the gene's key role in the immune system and suggests that treating patients with interferon, which are proteins released by immune cells to defend against viruses, may help manage disease in the early stages.

How useful are these findings?

The overarching aim of genome association studies is to not only correlate genes but also design treatments. For instance, the knowledge that interferons play a role in mediating a severe infection is already being used in drug therapies in the management of severe COVID. A study called the COVIFERON trial tested three kinds of interferon on the management of severe COVID but found no significant benefit in alleviating disease. Genomics studies reveal an association with certain conditions but don't necessarily explain how the genes direct the chain of chemical reactions that bring about an adverse outcome. But the knowledge of the gene helps to design targeted drugs. New technologies, such as CRISPR, allow genes to be tweaked or silenced and therefore this approach could be used to make new medicines. The GenOMICC study isn't the only one of its kind. Several consortia globally are working on identifying genes that may explain different disease outcomes.

Expected Question (Prelims Exams)

Q. Consider the following statements regarding GenOMICC, which is in news recently?

1. It is the study of genetic factors which leads to critical illnesses.
2. It brings together the clinicians and scientists from around the world for the study.

which of the above statements is/are correct?

(a) 1 only

(b) 2 only

(c) Both 1 and 2

(d) None

Expected Question (Mains Exams)

Q. What is GenOMICC? Discuss the nature and significance of the study of Genetic factors leading to critical illnesses. (250 Words)

Note: - The question of the main examination given for practice is designed keeping in mind the upcoming UPSC main examination. Therefore, to get an answer to this question, you can take the help of this source as well as other sources related to this topic.