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Study discovers single gene that controls 3 diseases

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Singapore: An international research consortium led by the Universitat AutÃ?Æ?Ã?Â²noma de Barcelona (UAB), Spain; Ciberer and the University of Wurzburg, Germany, discovered a gene that can cause three totally different diseases, depending on how it is altered. The findings have been published in the American Journal of Human Genetics.

The researchers used next-generation massive ultrasequencing techniques to identify the mutations that lead to Fanconi anemia. The team found that pathogenic mutations in the ERCC4 gene, which was previously known to cause xeroderma pigmentosum and a type of progeria, also led to Fanconi anemia.

The researchers have shown that the gene is involved in two DNA repair mechanisms by which cells maintain the stability of the genome, in such a way that the balance between these two repair systems will determine which of the three diseases the patient will contract. The researchers are now exploring the gene's role in breast cancer and ovarian cancer.

UAB professor Dr Jordi SurrallÃ?Æ?Ã?Â©s, said that, "This is a rather exceptional case, since there are few precedents of a single gene being involved in two independent physiological mechanisms and causing three clinically different diseases."