New disease gene discovery sheds light on cause of bone marrow failure

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The study, published in *The American Journal of Human Genetics*, detected and identified a new disease gene (ERCC6L2). In its normal form, the gene plays a key role in protecting DNA from damaging agents, but when the gene is mutated the cell is not able to protect itself in the normal way.

The research findings suggest that the gene defect and the subsequent

DNA damage was the underlying cause of bone marrow failure among the study participants.

Bone marrow failure is a term used for a group of life threatening disorders associated with an inability of the bone marrow to make an adequate number of mature blood cells.

Patients were recruited from all over the world to join an international bone marrow failure registry and researchers used new DNA sequencing technologies to study cases of bone marrow failure with similar clinical features. These included bone marrow failure associated with neurological abnormalities (learning defects and developmental delay), and patients whose parents were first cousins.

The findings mean it is now possible to carry out a reliable genetic test (including antenatal testing) in these families and get an accurate diagnosis. In the long term, with further research, the findings could lead to the development of new treatment for this specific gene defect.

Professor Inderjeet Dokal, Chair of Paediatrics and Child Health at Queen Mary University of London, comments:

"New DNA sequencing technology has enabled us to identify and define a new gene defect which causes a particular type of bone marrow failure. This is a promising finding which we hope one day could lead to finding an effective treatment for this type of gene defect. Clinicians treating patients with bone marrow failure should now include analysis for this gene in their investigation.

"Now we know this research technique works, we plan to carry out further studies to shed more light on the genetic basis of many other cases of bone marrow failure."

Medical Xpress

Provided by Queen Mary, University of London

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