

Key cause of bone marrow failure identified

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(Medical Xpress) -- Researchers funded by Leukaemia & Lymphoma Research and the Medical Research Council (MRC) have identified one of the first steps that can cause bone marrow failure and eventually lead to blood cancers. This discovery will open up a new strategy for treating these disorders and hopefully prevent some forms leukaemia, which affects up to 7,600 people a year in the UK.

The research, led by a team at the MRC Laboratory of Molecular Biology, uncovered that a defect in the process which controls how our cells produce proteins, co-ordinated by components of the cell called ribosomes, can lead to a bone marrow disorder called 'Shwachman-Diamond Syndrome' (SDS). In three out of every ten cases, SDS progresses to leukaemia. This discovery not only offers hope for new treatments for this and similar bone marrow illnesses, but also increases our understanding of how bone marrow failure can increase a person's chances of contracting blood cancers such as acute myeloid leukaemia and myelodysplastic syndrome (MDS).

These syndromes are some of the most widespread cancers of the blood, but little is known about the genetic basis of these disorders and there are currently no treatments other than bone marrow transplantation, which is available to only a small proportion of patients.

The findings are surprising and highly significant as they link a defective ribosome to bone marrow failure and leukaemia. They not only define the mechanism underlying a fundamental step in ribosome development, but also directly identify new approaches to treat the disease.

Dr Alan Warren, from the MRC Laboratory of Molecular Biology, said:

"We have defined SDS as a "ribosomopathy", linking the origins of this disorder to other forms of human bone marrow failure and cancer-related diseases in which ribosome dysfunction has been implicated. The next stage will be to focus on designing targeted treatments which exploit this new-found knowledge."

Dr David Grant, Scientific Director at Leukaemia & Lymphoma Research, said:

"This study will be hugely influential on future research into treatments for blood cancers that originate in the bone marrow. It could also lead to wider insights into how some leukaemias develop from this unexpected defect in protein production."

Collaborators with Alan's group included fellow researchers from the MRC Laboratory of Molecular Biology, Dr Andrew McKenzie, Dr Stefan Freund and researchers in Toronto and France.

The research was predominantly funded by Leukaemia & Lymphoma Research, the Medical Research Council and the Association for International Cancer Research as well as through a contribution from the charity Ted's Gang.

Provided by Medical Research Council

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