

The C-terminal domain of the Bloom syndrome DNA helicase is essential for genomic stability

## ABSTRACT

For this reason, the helicase activity and the C-terminal domain of BLM are important factors for maintaining genomic stability as measured by the sister chromatid exchange assay, and it seems that the amount of C-terminally abundant (GLD) localization of these molecules into the nucleolus by mutually agreed upon Cterminality appears to be more important for genomic instability than localized localisation in the nuclear bodies.

## INTRODUCTION

One of the most common genetic diseases in the world is the Bloom syndrome, a genetic disorder where a small proportion of affected individuals have normal chromosomes. The disease is associated with the deletion of one or more of the four exons of the mitochondrial DNA (mtDNA) gene, leading to the presence of an abnormal number of copies of the same gene in the nucleus of the cell.

The gene responsible for carrying out this cell division, the DNA helicase (DNase) gene, is located on chromosome 3 and involved in DNA replication. The DNase gene is a gene that has been shown to be involved in the normal functioning of the cell. It can also be involved in the maintenance of the cell cycle, in response to DNA damage. The number of copies of the DNase gene in the nucleus of the The RecQ family of DNA helicases encompasses BLM. Two reports have reported the development of Bloom syndrome, a rare autosomal recessive disorder characterized by genomic instability, immunodeficiency, small stature and chromatid instability in cells. Among the human genes in the RecQ family are RecQL/RecQL, WRN, RecQC5, and BLM. WRN is mutated in Werner syndrome, a disorder that affects premature aging. WS cells display genomic instability, while WRM encodes an exonuclease activity and shares many resemblances in vitro with BAM (breathers) and cancer-prone disorder. Previous work from this laboratory demonstrated that DNA helicase activity of B BLM is confined to specific areas of the nuclear body and its nucleolus. The BLM DNA helicase is found in two distinct nuclear structures in normal human cells, namely ND10 or PML nuclear bodies and the nucleolus. The NBs are dynamic PMI-dependent depots of various proteins disrupted upon viral infection and in some human malignancies. BS cells have a standardized RNA coding system that regulates the N-terminal and C- terminal domains for local regulation of cell-cycle homing (although their precise role remains unknown). The N

## CONCLUSION

Remarkable conclusions These data indicate that the helicase activity and C-terminal domain localization of BLM are crucial for maintaining genomic stability, as they directly direct BRM into nuclear bodies through delocalization.