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Short Communication

Human MMS21, also known as NSMCE2, Has New Functions in Development and Disease

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ABSTRACT

The transmission of entire genetic information by progeny cells requires chromosomal integrity during cell division. Errors in the systems that keep genomic stability during cell division can cause chromosomal aberrations during gametogenesis and development, which can show as disease phenotypes. Protein complexes involved in the structural maintenance of chromosomes (SMC) perform an important role. The Smc5/6 complex is required for chromosomal integrity, viability, and the repair of DNA double strand breaks and collapsed replication forks. This brief review focuses on hMMS21/NSMCE2, the human homolog of MMS21/NSE2, a non-smc component of the Smc5/6 complex with SUMO E3 ligase activity, and attempts to shed light on the function of hMMS21/NSMCE2 in human development and illness associated with NSMCE2 loss.

KEYWORDS: Down syndrome, trisomy 21, chromosome abnormality.

INTRODUCTION

Protein complexes that perform a dynamic scaolding function to shape chromosomes are known as Structural Maintenance of Chromosomes (SMC). Non-histone DNA linked proteins called SMC complexes are critical for higher order chromosomal structure. SMC proteins normally form V-shaped heterodimers with two ATP-binding head domains coupled to coiled regions and a hinge region connecting them. The SMC heterodimer interacts with additional subunits that are essential to the SMC complex's activity.

SMC complexes like condensin and cohesin govern several chromosomal structural features like condensation and sister chromatid cohesion. Furthermore, these SMC-complexes are engaged in a variety of chromosomal processes, including DNA double-strand break repair, replication, gene expression control, and so on. Deficiency in the genes encoding these complexes, notably cohesin and its loading factors, has been linked to serious developmental abnormalities in humans, which are now being researched. The Smc5/6 complex [1, 2, 3], a chromatin-associated, important complex whose Smc subunits are more closely linked to bacterial Smc-related proteins than the Smc1 or 3 or Smc2 or 4 heterodimers in cohesin and condensin, respectively. From microbes to humans, the intriguing Smc5/6 complex has been preserved through evolution. While its function is unknown, there is evidence that it is involved in a variety of chromosomal activities. The Smc5/6 complex is involved in both mitotic and meiotic proliferation, as well as the preservation of chromosomal integrity. It is involved in the rescue of collapsed replication forks, replication fork progression, DNA double strand break repair, and recombination intermediate resolution.

CONCLUSION

Finally, MMS21/NSMCE2 in mammals is required for proper development, chromosomal integrity, and tumour suppression in vivo, dentification of the complete spectrum of hMMS21/NSMCE2 sumoylation targets in human cells and determination of the functional significance of NSMCE2-mediated sumoylation in various cell types and developmental lineages may give further insights into comprehending all of the dall the defective phenotypes associated with NSMCE2 dysfunction in humans.

REFERENCES

1. Dierssen M, Herault Y, Estivill X (2009) Aneuploidy: from a physiological mechanism of variance to Down syndrome. Physiol Rev 89: 887-920.
2. Cook EH Jr, Scherer SW (2008) Copy-number variations associated with neuropsychiatric conditions. Nature 455: 919-923.
3. Veitia RA, Potier MC2 (2015) Gene dosage imbalances: action, reaction, and models. Trends Biochem Sci 40: 309-317.

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