

New Functions in Down Syndrome

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INTRODUCTION

Protein complexes that perform a dynamic scaffolding 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 [1].

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 [2], 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 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 [3].

CONCLUSION

Finally, MMS21/NSMCE2 in mammals is required for proper development, chromosomal integrity, and tumour suppression *in vivo*, identification 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 different defective phenotypes associated with NSMCE2 dysfunction in humans.

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Received date: July 09, 2021; Accepted date: July 23, 2021; Published date: July 30, 2021

Citation: Sethaphanich N (2021) New Functions in Down Syndrome. *J Down Syndr Chr Abnorm*, 7: 174. doi: 10.4172/2472-1115.21.7.174

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