Understanding the cause of ataxia in the DNA repair deficiency syndrome Ataxia Telangiectasia: a possible link between DNA damage and proteostasis.

https://neurodegenerationresearch.eu/survey/understanding-the-cause-of-ataxia-in-the-dna-repair-deficiency-syndrome-ataxia-telangiectasia-a-possible-link-between-dna-damage-and-proteostasis/

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Funder

Hersenstichting | Dutch Brain Foundation

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The Netherlands

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Understanding the cause of ataxia in the DNA repair deficiency syndrome Ataxia Telangiectasia: a possible link between DNA damage and proteostasis.

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Research Abstract
‘The human syndrome Ataxia Telangiectasia (AT) is caused by a mutation in the ATM gene.

Patients with this syndrome have problems with their immune system and a greater risk of cancer. This is explained by the fact that the ATM protein has an important function in repairing DNA damage. However, in patients (and several other syndromes with DNA damage repair problems), there are also neurodegeneration (ataxia), but what is underlying here is unclear. The purpose of this research is to better understand why problems with DNA damage repair lead to neurodegeneration. It is known that most neurodegenerative diseases are caused by accumulation of wrong folded proteins, which then merge (aggregate) and disrupt the function of cells, eventually leading to cell death. Preliminary neurons appear extremely sensitive to this protein aggregation. The key hypothesis I want to test is whether or not (well) recovered DNA damage can also disrupt the protein balance in the cell. The idea is that since DNA can be read to make a protein, errors in the DNA can lead to less stable proteins (proteins with “errors”). These less stable proteins can aggregate and ultimately lead to neurodegeneration.’

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