

ATAXIA-TELANGIECTASIA

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ATM GENE PRODUCT

- ATM gene produces a protein found in the nucleus of all cells
- protein plays diverse roles including:
 - cell growth
 - cell division
 - development and activity of nervous system
 - development and activity of immune system
 - recognizes damaged DNA
 - coordinates repair of damaged DNA
- consequences of mutated ATM protein:
 - abnormal cell growth and division → cancerous tumors
 - premature cell death
 - altered brain cerebellar function → diminished coordination of movement (ataxia)
 - compromised immune system
 - breakage of chromosomes

