

Chapter 18
Web Text Box 3

What is Ataxia Telangiectasia?

On book page 306 we describe how the protein kinase ATM acts to prevent cells passing either the G2/M and G1/S cell cycle control points. The abbreviation ATM stands for ataxia telangiectasia mutated, because mutation of this gene results in the rare, progressive, neurodegenerative childhood disease ataxia telangiectasia. As they start to walk children exhibit poor balance (ataxia) and slurred speech together with "spider" veins in their eyes, ears and cheeks (telangiectasia). They also show premature graying of the hair and delayed physical and sexual development. Because of the involvement of ATM in the DNA damage response AT children show extreme sensitivity to ionizing radiation. Normally harmless doses of X-rays, such as those administered by a dentist, cause severe burns to an AT child, 30% of whom develop lymphoid cancers.

The charity supporting families in the USA with ataxia telangiectasia is the National Ataxia Foundation <http://www.ataxia.org/>. See also the British ataxia telangiectasia society at <http://www.atsociety.org.uk/>.