

## Management of Individuals With a Mutation in the Ataxia Telangiectasia Mutated Gene

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Advances in genetic testing have led to the identification of multiple genes associated with a hereditary risk for developing breast and other cancers. One such gene is the ataxia telangiectasia mutated (*ATM*) gene, which is available on many genetic panels offered to individuals with suspected hereditary risk. Genetic testing can often lead to improved understanding and clarification of risk for developing cancer, as well as allow affected individuals to make informed choices about management, including the adoption of primary prevention strategies and more aggressive screening than typically recommended in the general population. This article provides an overview of the role of mutations in the *ATM* gene in developing malignancies, along with emerging research on treatment implications based on genetic testing results.

### Physiologic Function

The *ATM* gene is located on the long (q) arm of chromosome 11 between positions 22 and 23, spanning from base pair 108,222,499 to base pair 108,369,101 (Genetics Home Reference, 2015). The *ATM* gene is large; with 63 exons containing about 150 kb of genomic DNA, analyzing the gene is technologically difficult (Graña et al., 2011; Mitui et al., 2009). In addition, the *ATM* gene provides the code to

produce a protein that functions in the nucleus of the cell (Genetics Home Reference, 2015).

The function of the *ATM* protein is to regulate the rate at which cells grow and divide, as well as to promote the normal development and activity of the nervous and immune systems. This protein also plays an important role in recognizing damaged or broken DNA strands and facilitating DNA repair by activating enzymes to repair the damaged DNA strands. The *ATM* gene is considered to be a major guardian of genomic stability (Shiloh, 2014).

The impact of mutations in the *ATM* gene depends on the number of alleles affected. When an individual is homozygous (both alleles are affected) for a mutation in the *ATM* gene, the person will develop ataxia telangiectasia (AT), a rare autosomal recessive disorder (Lindor, McMaster, Lindor, & Greene, 2008). Individuals who are heterozygous (one copy is affected) are at increased risk for developing several malignancies, including breast, colon, stomach, bladder, pancreatic, lung, and ovarian cancers (Gatti, 2010).

### Clinical Manifestations

**Homozygous:** AT occurs because of a germline biallelic (both copies affected) mutation in 1 out of every 40,000 to 100,000 live births (Gatti, 2010). In individuals with suspected AT, germline sequence analysis of