PublisherInfo		
PublisherName	:	BioMed Central
PublisherLocation	:	London
PublisherImprintName	:	BioMed Central

ATM splicing defect

ArticleInfo		
ArticleID	:	4422
ArticleDOI	:	10.1186/gb-spotlight-20020314-01
ArticleCitationID	:	spotlight-20020314-01
ArticleSequenceNumber	:	88
ArticleCategory	:	Research news
ArticleFirstPage	:	1
ArticleLastPage	:	2
ArticleHistory	:	RegistrationDate : 2002–3–14 OnlineDate : 2002–3–14
ArticleCopyright	:	BioMed Central Ltd2002
ArticleGrants	:	
ArticleContext	:	130593311

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Individuals with mutations in the *ATM* gene develop ataxia-telangiectasia, a neurodegenrative disorder characterized by immunological defects and cancer predisposition. In an Advanced Online Publication from Nature Genetics, Pagani *et al.* describe a new kind of *ATM* mutation that leads to an unusual splicing defect (11 March 2002, DOI:10.1038/ng858). The mutant *ATM* allele contains a four-nucleotide deletion (GTAA) within intron 20. This deletion results in the inclusion of a 65 nucleotide 'cryptic exon' in the *ATM* mRNA. The ATM sequence, termed intron-splicing processing element (ISPE), is complementary to U1 snRNA. Experiments with a hybrid minigene confirmed the importance of the ISPE sequence; and interaction with U1 snRNA affected the efficiency of intron removal. Introduction of the ISPE sequence into a different genomic context, exon 9 of the cystic fibrosis transmembrane regulator (*CFTR*) gene, caused exon skipping and splicing defects.

References

1. The genetic defect in ataxia-telangiectasia.

2. *Nature Genetics*, [http://www.nature.com/ng/]