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ATR mutation

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Seckel syndrome is an autosomal recessive disorder involving dwarfism and mental retardation. In an Advance Online Publication in Nature Genetics O'Driscoll *et al.* report the discovery of a mutation in the DNA-repair kinase ATR, in Seckel syndrome patients (*Nature Genetics*, 17 March 2003, doi:10.1038/ng1129). The ATR(ataxia-telangiectasia and Rad3-related) gene mapped to chromosome 3q22.1-q24, which had been genetically linked to Seckel syndrome. Fibroblasts from patients had impaired ATR function and response to ultraviolet radiation. O'Driscoll *et al.*identified a synonymous mutation that affects ATR splicing, and that causes reduced ATR protein levels. This is the first report linking ATR signalling to a clinical disorder.

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