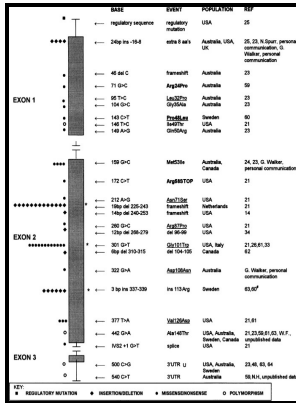


# Role of germline CDKN2A mutations in the development of melanoma and other malignancies

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## Germline CDKN2A mutation implicated in predisposition to multiple myeloma

Transient transfection was carried out with Lipofectamine 2000 Invitrogen. One of the gene transcripts ARF functions as a stabiliser of p53 through interaction with E3 ubiquitin protein ligase MDM2, thereby enhancing p53-dependent transactivation and apoptosis. E1013X in BRCA1, 7 of the frameshift p.

## Role of CDKN2A Mutations and Other Relevant Genes in Melanoma Predisposition

Tumors diagnosed before or after the study period were not included in the analysis. The possible role of cyclin-dependent kinase inhibitor CDKI genes was raised by recognition of cyclin D1 as a parathyroid oncogene, identification of rare germline mutations in CDKI genes in patients with multiple endocrine neoplasia type 1; that in rodents, mutation in Cdkn1b caused parathyroid tumors; and subsequently through identification of rare predisposing germline sequence variants and somatic mutation of CDKN1B, encoding p27 kip1, in sporadic human parathyroid adenoma. The p16 INK4A protein attaches binds to two other proteins called CDK4 and CDK6.

## New founder germline mutations of CDKN2A in melanoma

The aim of this study was to evaluate survival from all causes, from melanoma alone, and from nonmelanoma cancers in familial melanoma cases with mutations in the CDKN2A gene, compared with familial and sporadic cases without such mutations. Risk of solid tumors and myeloid hematological malignancies among first-degree relatives of patients with monoclonal gammopathy of undetermined significance.

## Constitutional mutation in CDKN2A is associated with long term survivorship in multiple myeloma: a case report

To compare the distribution of types of mutations between the multiple cancer and the UK10K control cohort, we used a Monte Carlo version of a chi-squared test with 1,000,000 randomisations. As murine plasmacytoma corresponds to multiple myeloma in humans, there has been considerable interest in the genetic basis for susceptibility to this disease.

## CDKN2A gene: MedlinePlus Genetics

A variant in ASXL1 p.

### **Role of CDKN2A Mutations and Other Relevant Genes in Melanoma Predisposition**

This includes 65 autosomal dominant cancer predisposition genes, 31 autosomal recessive cancer predisposition genes, 23 genes encoding tyrosine kinases, 58 tumour suppressor genes, 232 cancer associated genes and 116 DNA repair genes. More generally, evidence that CDKIs are authentic drivers in human tumorigenesis has been sparse, despite the appeal of their candidacy and efforts to mimic their actions therapeutically.

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