

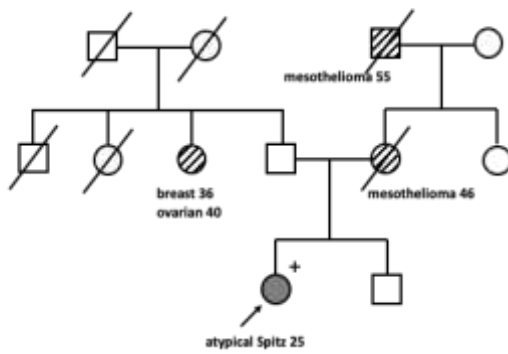
Article

# Insights into genetic susceptibility to melanoma by gene panel testing: potential pathogenic variants in *ACD*, *ATM*, *BAP1*, and *POT1*

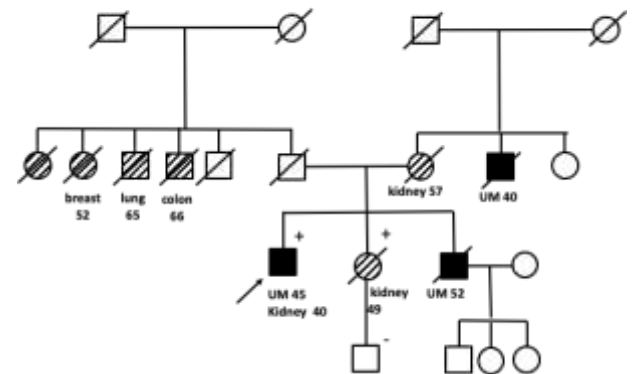
Supplementary Material

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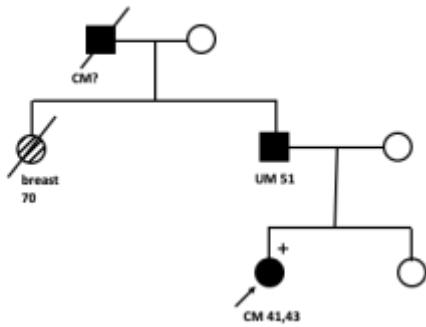
BAP1:c.327\_328insGA;p.(Pro110AspfsTer4)



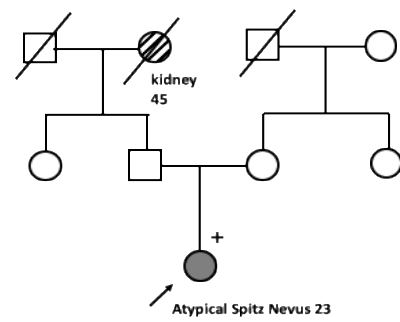
BAP1: c.639dupT;p.(Ile214TyrfstsTer29)



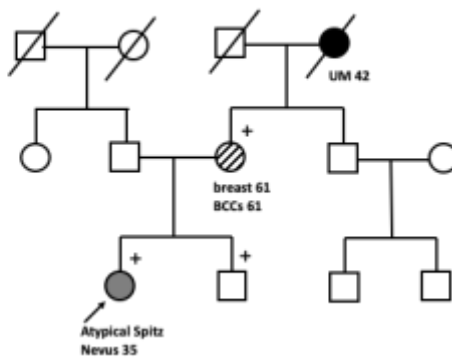
BAP1: c.1337delA;p.(Asn446ThrfsTer125)



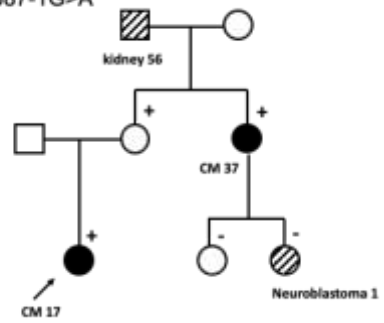
BAP1: c.1777dupC;p.(Gln593ProfsTer50)



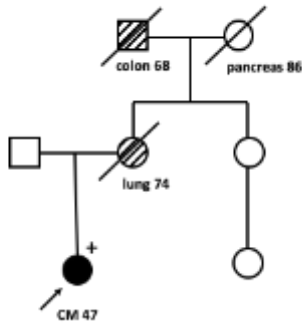
BAP1: c.1939G>T; p.(Glu647Ter)



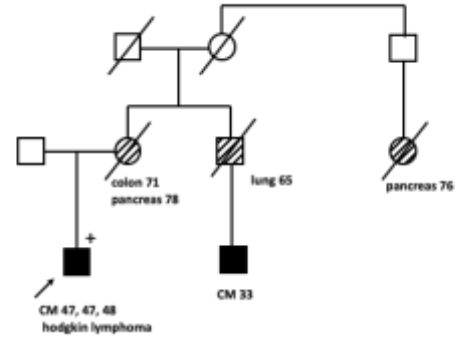
POT1: c.1687-1G>A



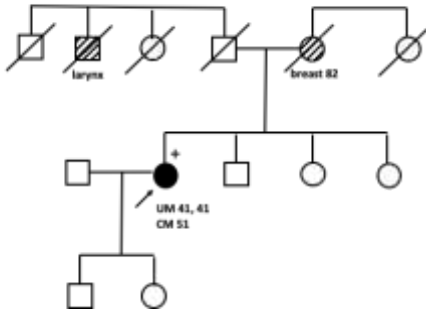
**ATM: c.3275C>A; p.Ser1092Ter**



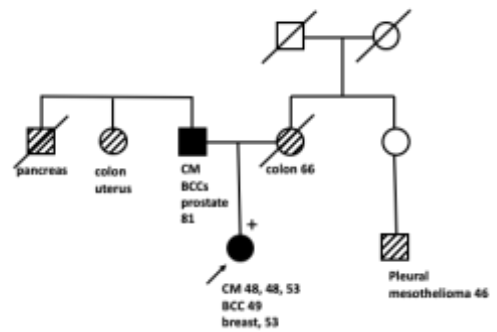
**ATM : c.4451delT; p.M1484fs**



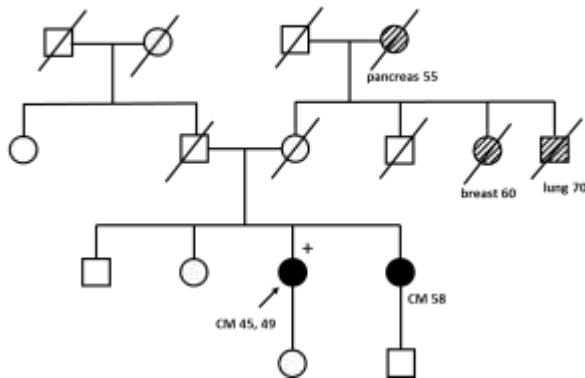
**ATM: c.5979\_5983delTAAAG; p.(Ser1993ArgfsTer23)**



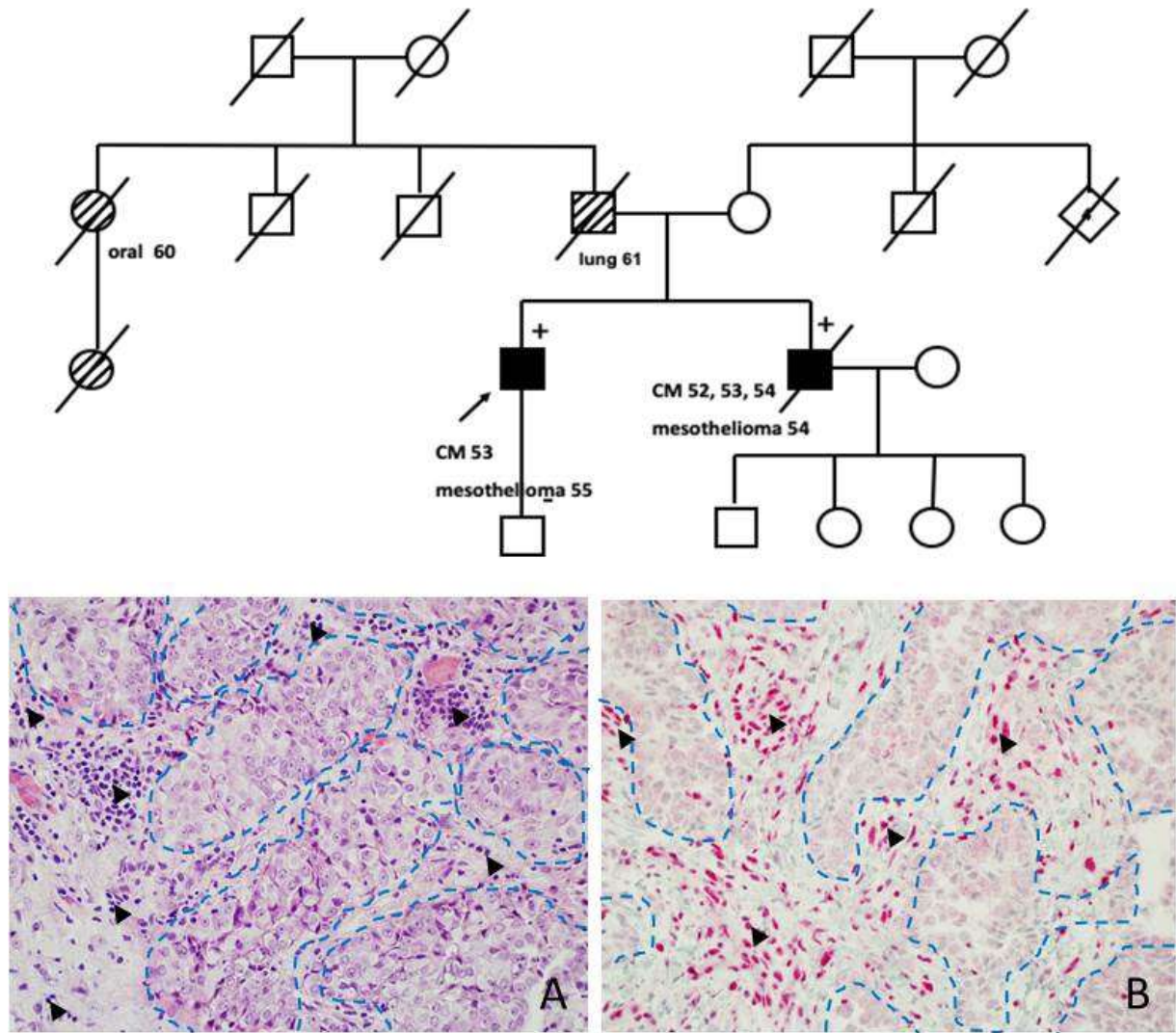
**ATM: c.8319\_8323dupTGTC; p.(Pro2775LeufsTer33)**



**ACD: c.866\_867delCT; p.(Pro289ArgfsTer28)**



**Figure S1.** Pedigree diagrams from families carrying loss of function variants in *BAP1*, *POT1*, *ATM* and *ACD* genes. Dark symbol: cutaneous melanoma (CM), or uveal melanoma (UM). Cancer type and age at diagnosis is indicated under each symbol. The proband is indicated by an arrow.



**Figure S2.** Pedigree diagram from the family with the rare *c.799\_800delCA BAP1* germline variant. The proband developed CM and was diagnosed with mesothelioma during mutation-specific follow-up. His brother had previously developed 3 CMs and mesothelioma. (A) Haematoxylin and Eosin stained section of mesothelioma from a pleural biopsy (magnification  $\times 40$ ) from the proband’s brother carrying the *c.799\_800delCA BAP1* germline variant. Within the dotted blue outline are neoplastic cells; the black arrow heads point to tumor infiltrating lymphocytes. (B) Section from the mesothelioma in (A) (magnification  $\times 40$ ) immunostained for BAP1 protein. Nuclear loss of BAP1 expression is seen in neoplastic cells within the dotted lines while the non-neoplastic lymphoid cell nuclei which serve as a control (showing no loss of BAP1 expression) stain in red (black arrow heads). Dark symbol: cutaneous melanoma (CM). Cancer type and age at diagnosis is indicated under each symbol. The black arrow in the pedigree points to the proband.

