

#AAD2019

Highlights
AEDV

IN 77TH AAD CONGRESS

1-5 MARCH 2019

★ WASHINGTON ★

Scientific Initiative of:



Sponsored by:



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Genes & Melanoma
Dr. David Moreno Ramírez

Scientific Initiative of:



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MELANOMA GENETICS

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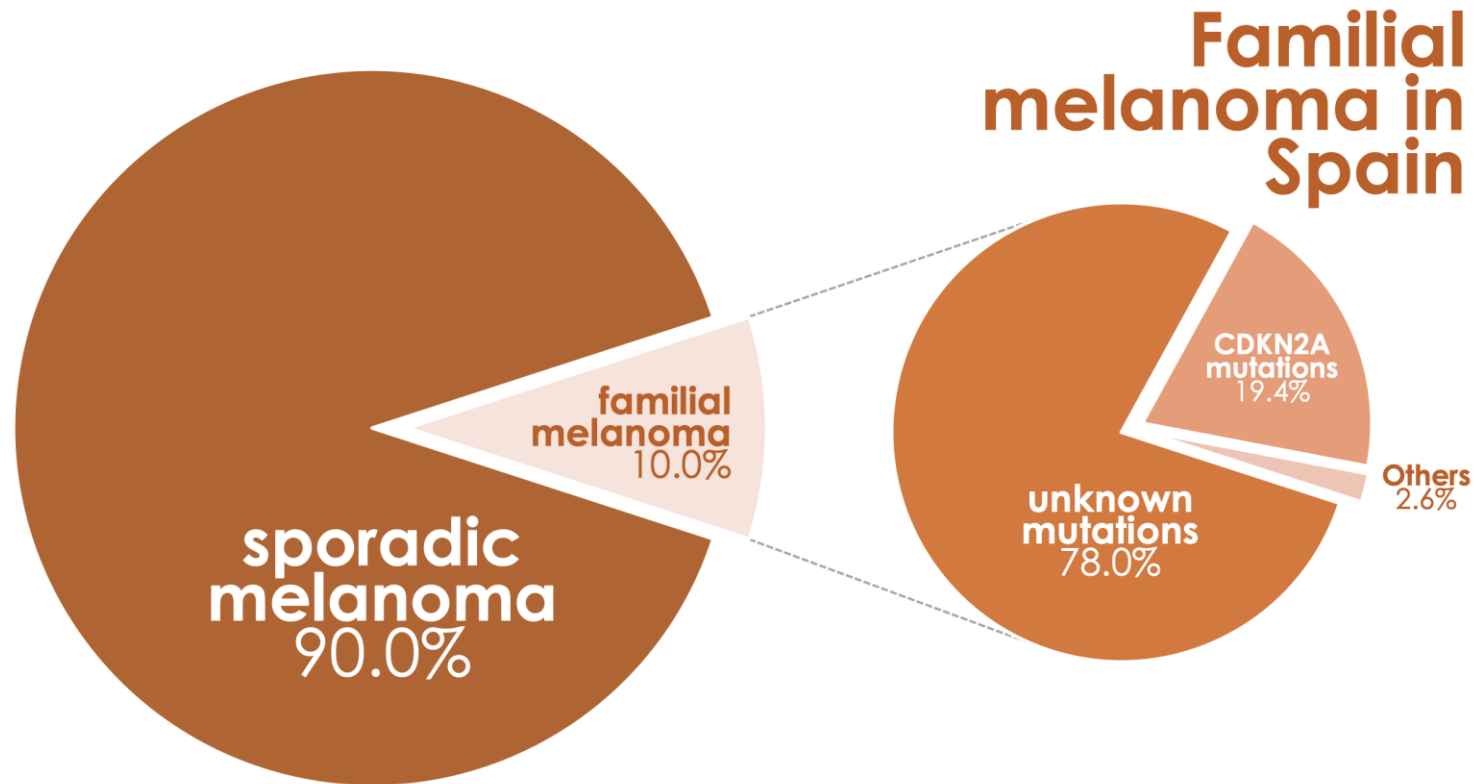
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Germline mutations

Somatic mutations

Mutations in germ cells DNA	Mutations in tumoral cells DNA
Familial melanoma Tumor predisposition syndrome	Tumor promotion Therapeutic targets Outcome information
Susceptibility genes	Driver mutations Tumor promoting proteins
CDKN2A CDK4 BAP-1 MCR1 POT1	BRAF NRAS TERT MTIF PTEN

CDKN2A GERMLINE MUTATIONS



Potrony M. Update in genetic susceptibility in melanoma
Ann Transl Med 2015

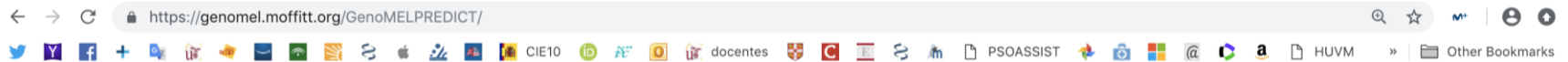
CDKN2A GERMLINE MUTATIONS

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Q1. How old were you when you were first diagnosed with melanoma?



Q2. How many primary melanomas have you had?



Q3. How many other members of your family have been diagnosed with melanoma?



Q4. Have you or any of your family members been diagnosed with pancreatic cancer?

Yes

Update

Clear

Based on the MELPREDICT model (Q1, Q2 & Q3), the most likely probability that you carry a mutation in *CDKN2A* is 2.8%. This estimate falls within a range of 2% to 3.7%.

Based on the 3-factor GenoMELPREDICT model (Q1, Q2 & Q3), the most likely probability that you carry a mutation in *CDKN2A* is 25.2%. This estimate falls within a range of 19.8% to 31.6%.

Based on the 4-factor GenoMELPREDICT model (Q1, Q2, Q3 & Q4), the most likely probability that you carry a mutation in *CDKN2A* is 47%. This estimate falls within a range of 40.3% to 53.8%.

BAP-1 TUMOR PREDISPOSITION SYNDROME

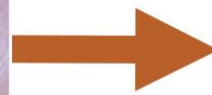
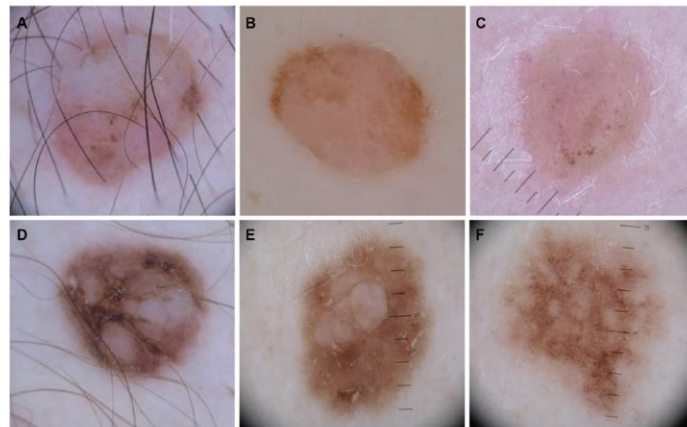
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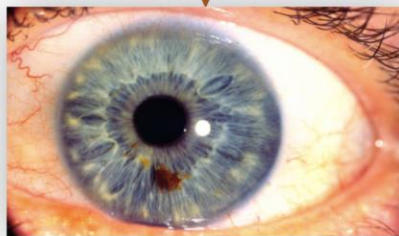


Biopsy

Confirm BAP1
inactivated tumor

BAP1 testing?

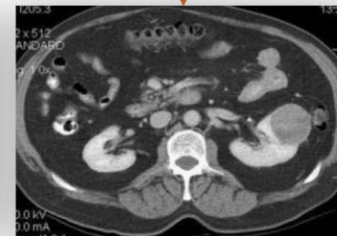
**Cancer
screening?**



**Ophthalmologic
examination
yearly**



Imaging



**Total skin examination
every 6 months**

TERT -TELOMERASE REVERSE TRANSCRIPTASE- PROMOTER MUTATIONS

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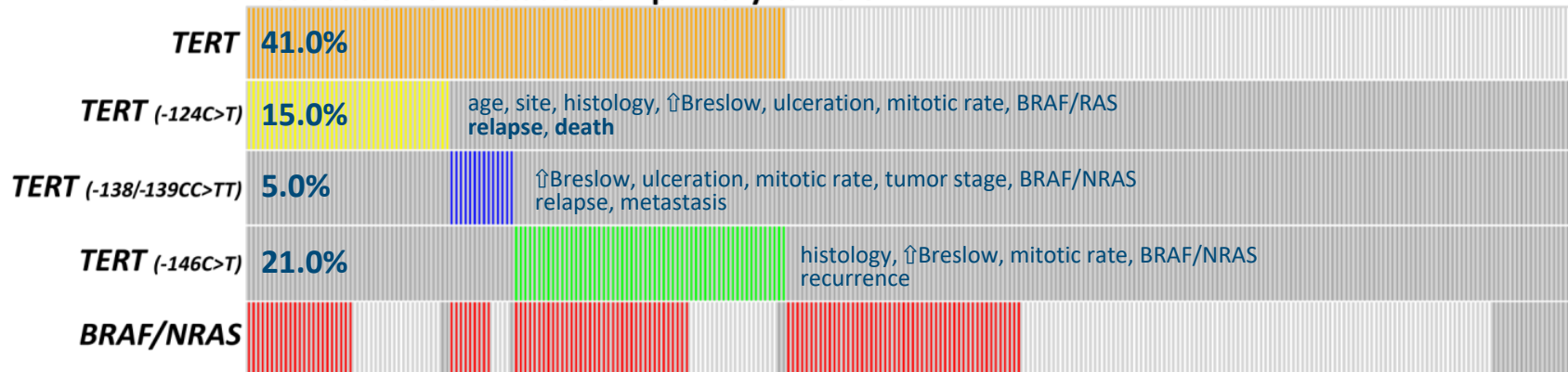
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Distribution of mutations in primary tumors



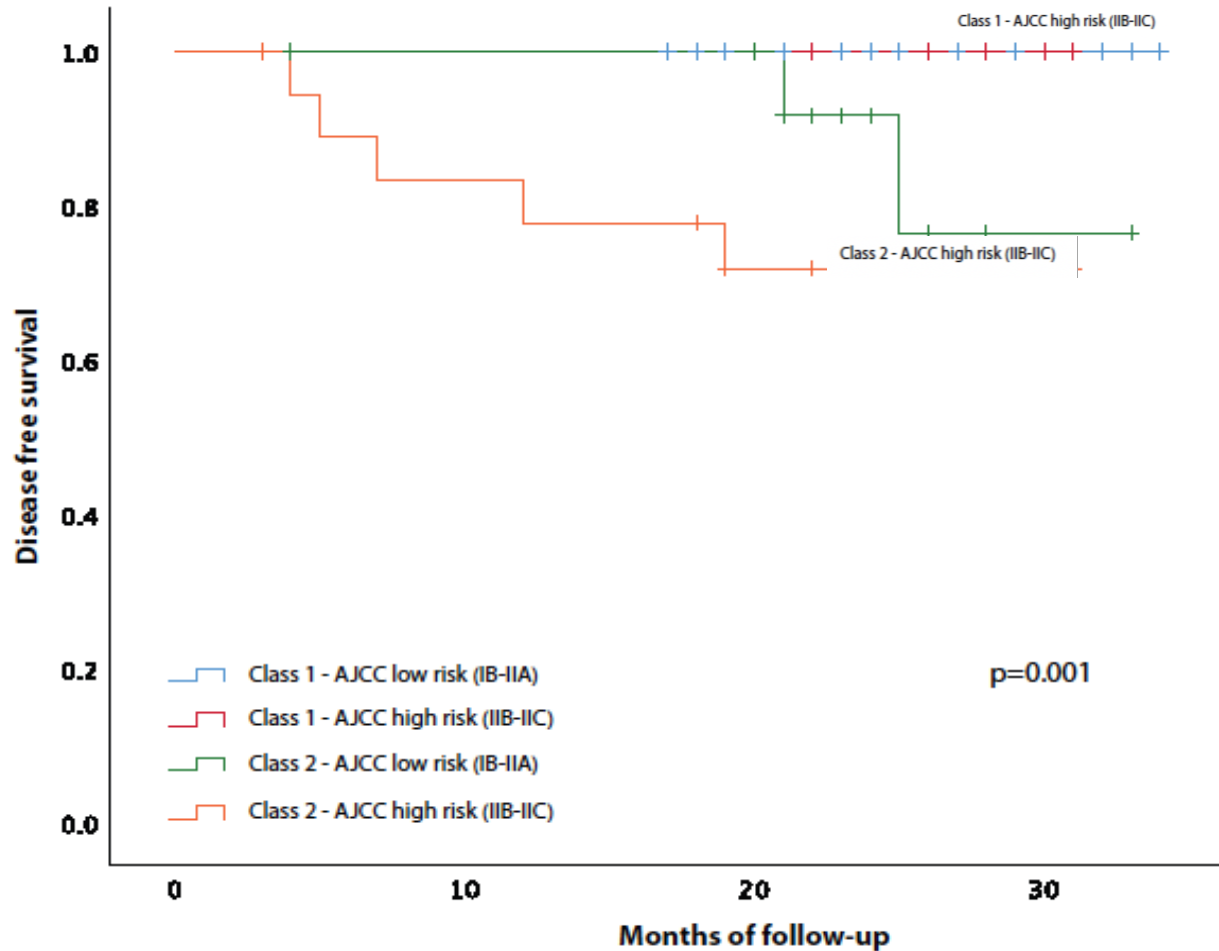
31-GEN EXPRESSION PROFILE



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Podlipnik et al.
Early outcome of a **31-gene expression profile** test in 86 AJCC stage IB-II melanoma patients. A prospective multicentre cohort study. J EADV 2019

TO TAKE HOME...

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- GenoMELPREDICT CDKN2A testing?
- Take care of BAP1 inactivated nevus!!
- TERT mutations as survival predictor
- 31-GEP available but not yet fully incorporated

