SNPixel

Find the right pixel in the genome - NF2 case study

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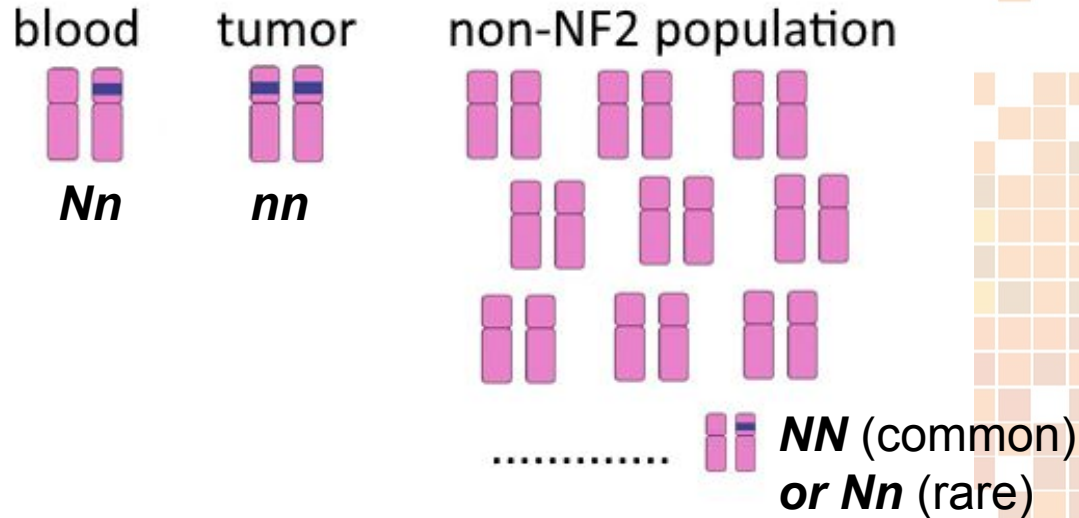Selina Kim

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Annotating the SNPs

Predicts the effects of variants on genes based on amino acid changes

Predict the effects of variants based on regulatory elements

Screening candidate variant based on an inherited & a sporadic mutation hypothesis and variant frequency in populations
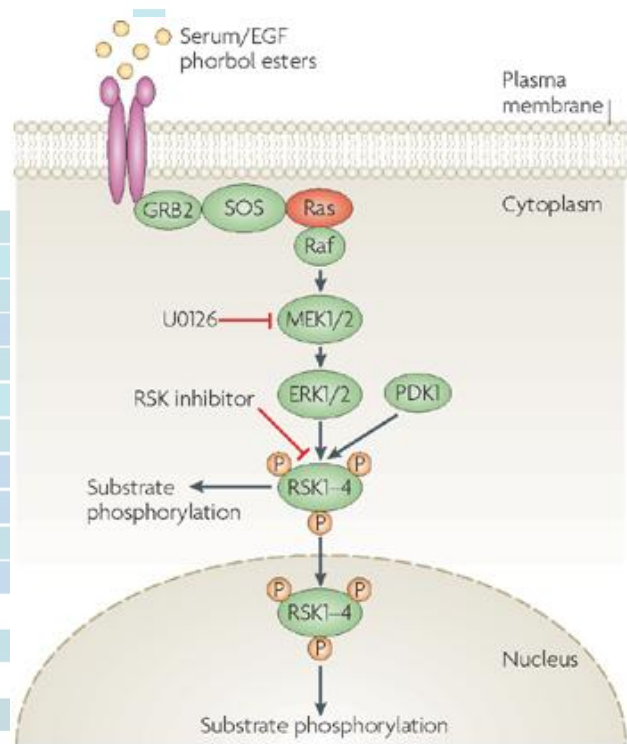
blood   tumor   non-NF2 population

Nn   *nn*

....   *NN* (common)
or Nn (rare)

Sporadic mutation in NF2 (or any gene)
-   Blood: inherited mutation (heterozygote, *Nn*)
-   Tumor: +spontaneous mutation (homozygote, *nn*)
-   Others: almost no mutation (homozygote, *NN*)

Demo

RSK activation pathway

RPS6KA2

6	166816025	-------------	CCTCTCTCTCT	C	intron_variant	MODIFIER

RPS6KA6 (only sporadic mutation)

X	84059139	rs6623143	C	T	3_prime_UTR_variant	MODIFIER
X	84059146	rs6623144	A	C	3_prime_UTR_variant	MODIFIER
X	84059157	rs6623146	A	C	3_prime_UTR_variant	MODIFIER
X	84062780	rs6623149	T	C	3_prime_UTR_variant	MODIFIER
X	84062783	rs6623150	C	T	3_prime_UTR_variant	MODIFIER
X	84072592	--------------	T	G	intron_variant	MODIFIER
X	84073096	--------------	G	T	intron_variant	MODIFIER

MAPK1

22	21784594	--------------	CCTCTTTCTTTTTT	C	intron_variant	MODIFIER

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Genome editing for NF2 modeling

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