

# SNPixel

Find the right pixel in the genome - NF2 case study

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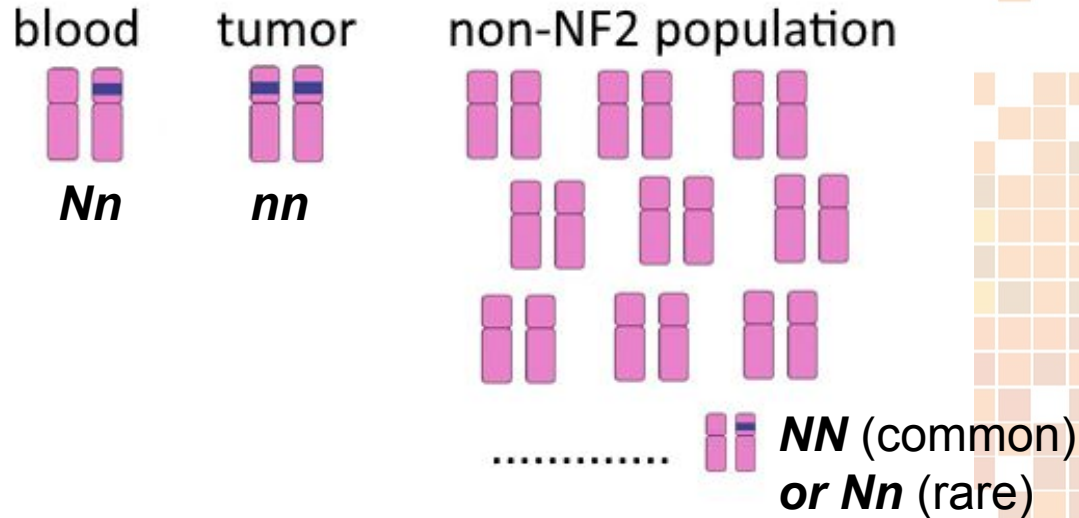
recombinetics

# 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

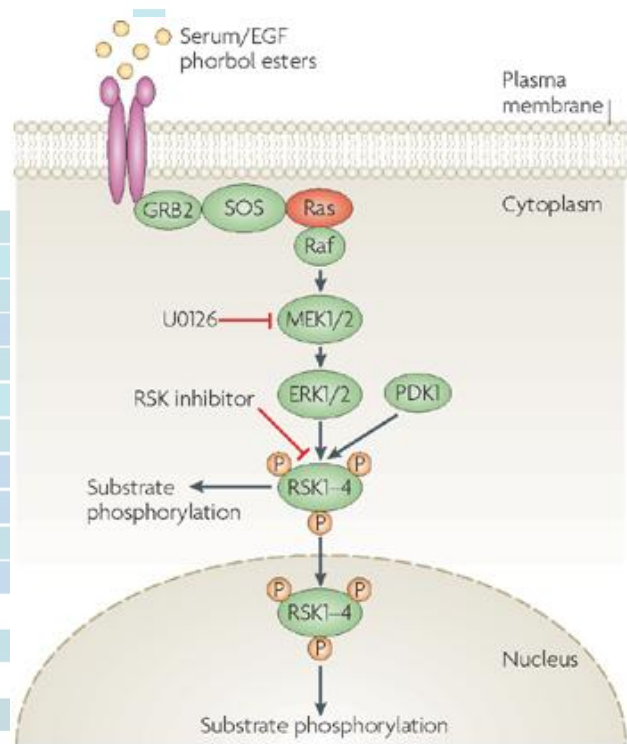


## Sporadic mutation in NF2 (or any gene)

- Blood: inherited mutation (heterozygote, *Nn*)
- Tumor: +spontaneous mutation (homozygote, *nn*)
- Others: almost no mutation (homozygote, *NN*)

The background is a large, pixelated graphic. It features a horizontal color gradient: blue on the left, transitioning through green and yellow in the center, and ending in red on the right. The pixels are arranged in a grid-like pattern, with some areas being more densely packed than others. The word "Demo" is centered in the middle of the image, overlaid on the yellow and green pixelated area.

Demo



Nature Reviews | Molecular Cell Biology

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 ----- CCTCTTTCTTTTT C intron\_variant MODIFIER

ss#	Sample Ascertainment			Genotypes		Alleles	
	Population	Individual Group	Chrom. Sample Cnt.	Source	HWP	C	T
ss1555147700	EAS		1008	AF		1.00000000	
	EUR		1006	AF		0.96319997	0.03680000
	AFR		1322	AF		0.94550002	0.05450000
	AMR		694	AF		0.97259998	0.02740000
	SAS		978	AF		0.99690002	0.00310000

# Genome editing for NF2 modeling



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