Effect of Mutation Types

Descriptions of Our Dataset Columns

Patient identifier

barcode	project_id	sample_type	primary_diagnosis	TP53_substitution_category	TP53_expr
TCGA-EB-A4IS-01A-21R-A266-07	TCGA-SKCM	Primary Tumor	Malignant melanoma, NOS	wild-type	35.581636
TCGA-EB-A3HV-01A-11R-A21D-07	TCGA-SKCM	Primary Tumor	Nodular melanoma	wild-type	41.618961
TCGA-BF-A3DJ-01A-11R-A20F-07	TCGA-SKCM	Primary Tumor	Malignant melanoma, NOS	wild-type	21.042620
TCGA-EB-A41B-01A-11R-A24X-07	TCGA-SKCM	Primary Tumor	Nodular melanoma	wild-type	10.771012
TCGA-EB-A42Z-01A-12R-A24X-07	TCGA-SKCM	Primary Tumor	Malignant melanoma, NOS	wild-type	35.305502
TCGA-EB-A431-01A-11R-A266-07	TCGA-SKCM	Primary Tumor	Nodular melanoma	nonsense	3.677341

Research Question-

How does the presence of a missense or nonsense mutation type affect the expression level of the gene compared to wild-type for CDKN2A, PTEN, RB1, TP53, ARID2, and NF1?

Graph Key and Vocab

Missense-

A mutation that switches amino acids

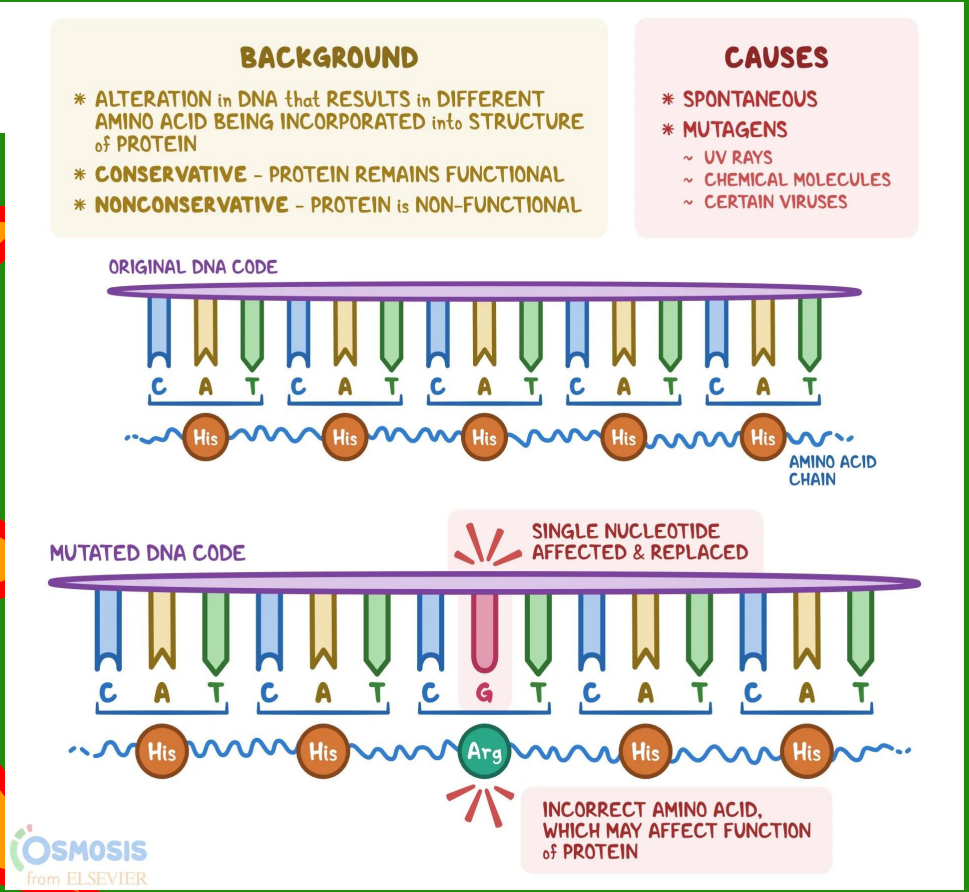
Amino acids- A group of 3 letters of DNA

Nonsense-

A mutation in the DNA that causes a stop order

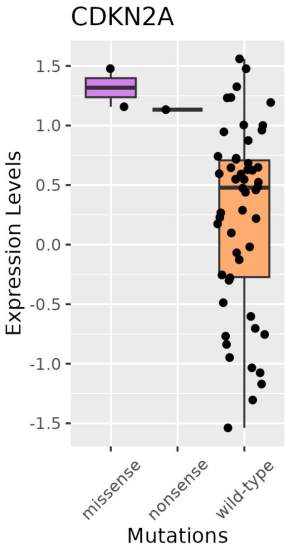
Wildtype-

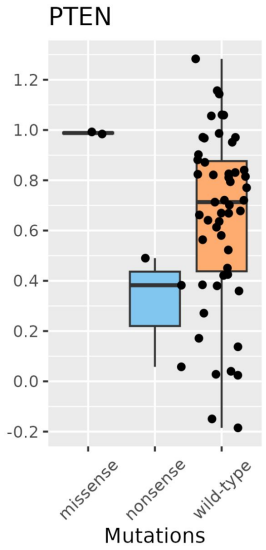
A mutation that happens naturally. It can be either good, bad or neutral.

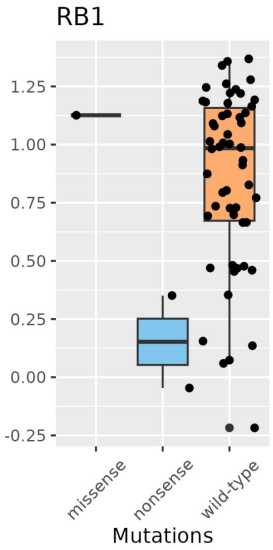


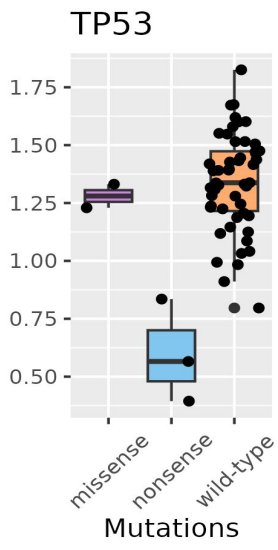
What does the data tell us?

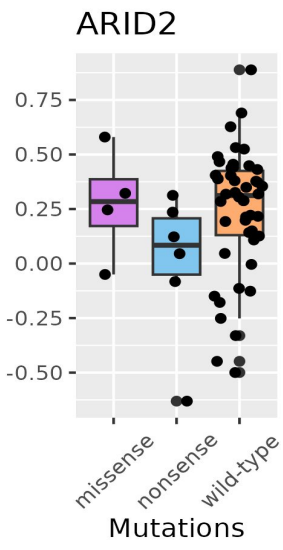
1 Dot = 1 patient

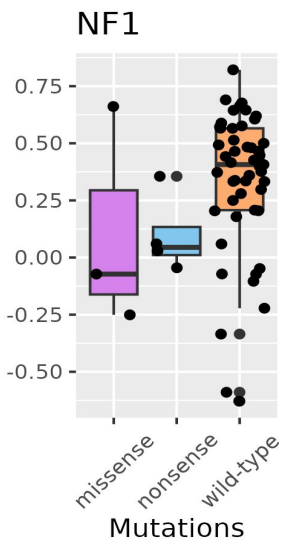












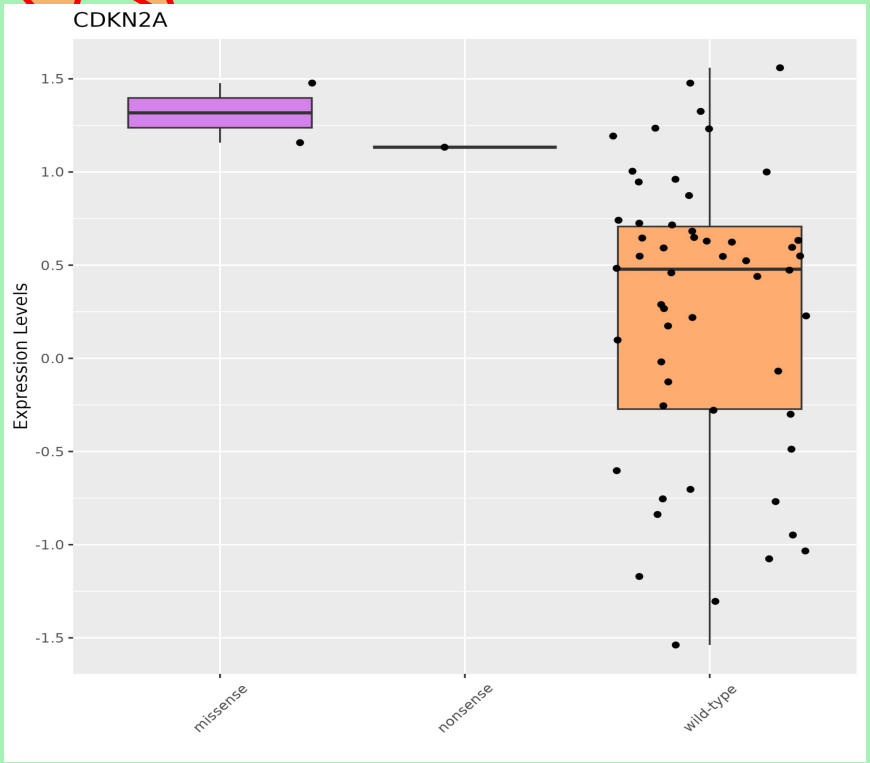
- Throughout all genes, wild-type has the most data points/patients.
- Based on the data charts missense has the least amount of patients.
- Throughout all genes (besides CDKN2A), nonsense mutations consistently holds a lower median expression level than wild-types

CDKN2A

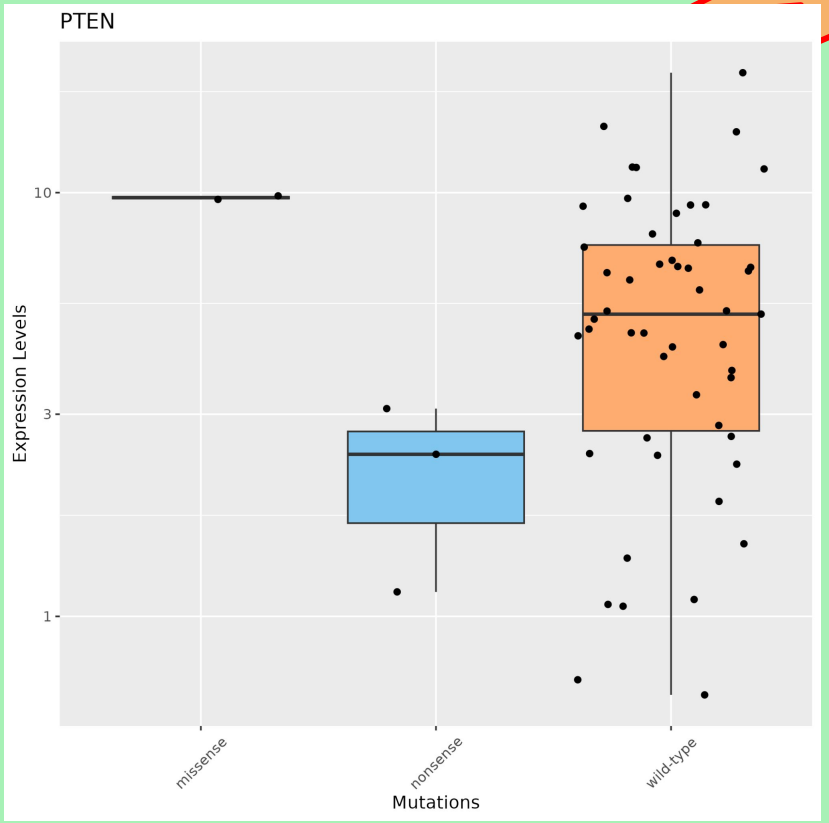
- The CDKN2A gene provides instructions for making several proteins. The two best studied ones, function as tumor suppressors.

- A mutation in this gene causes cells to grow and divide too quickly and may keep cells from undergoing apoptosis (“Cell death”)

- CDKN2A is altered in 10.19% of all cancers.



PTEN



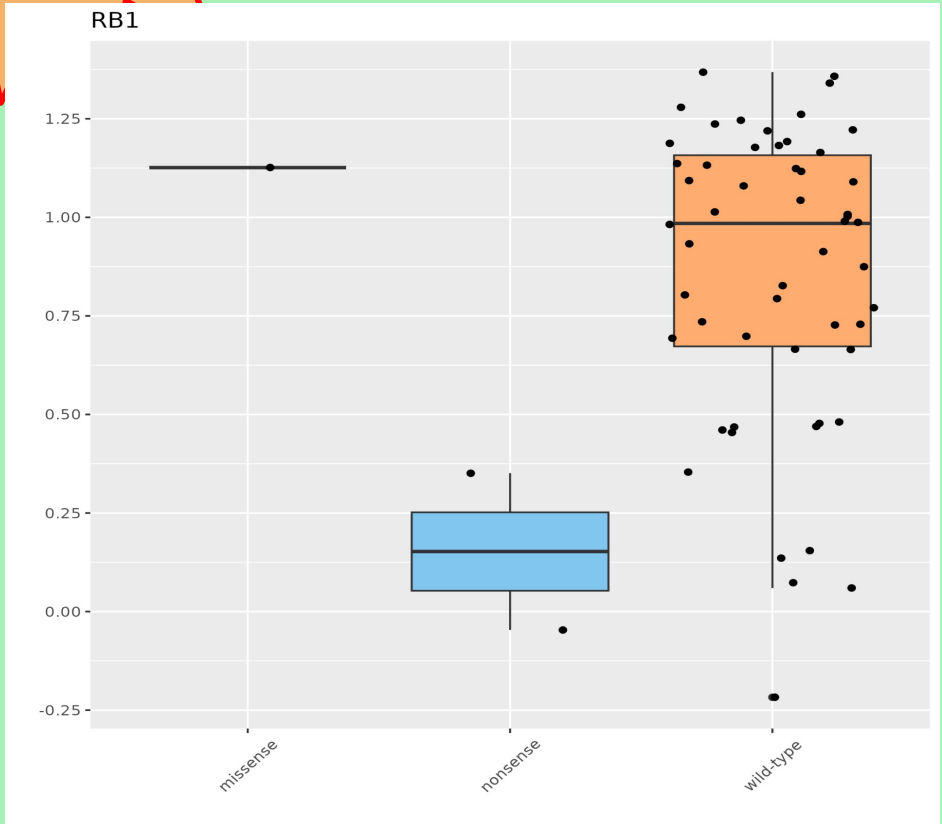
- PTEN gene gives instructions to make an enzyme, this enzyme acts like a tumor suppressor
- Mutations in PTEN would either reduce or eliminate the function of the tumor suppressor
- Nonsense mutations are most common in this gene

RB1

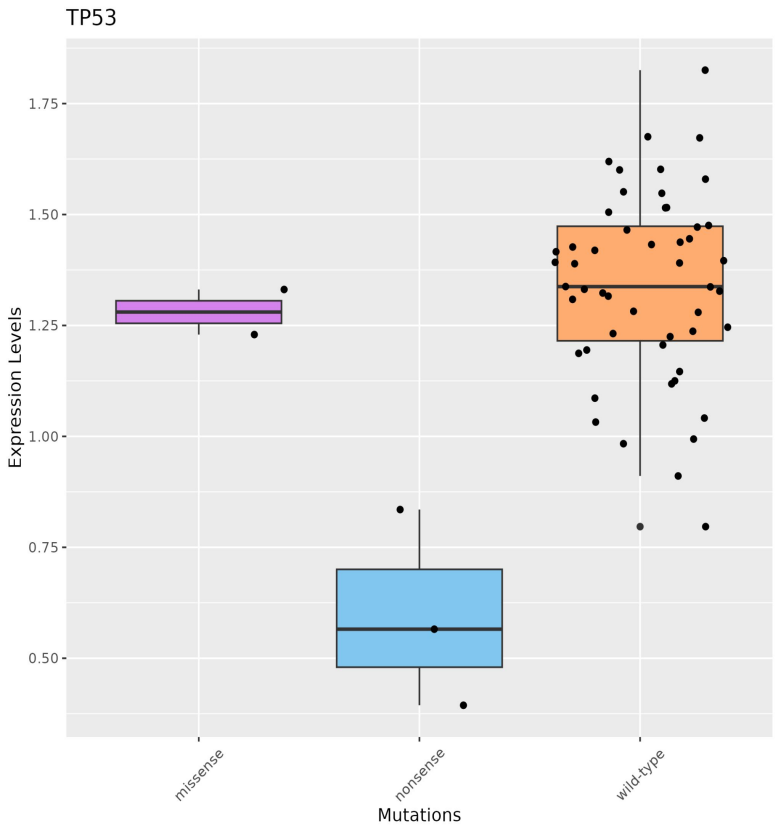
- Creates the pRB protein, which regulates cell division and growth. The pRB protein also prevents other proteins from triggering DNA replications

- A mutated RB1 gene can cause several cancers.

- Wild-type is the most common and maintains a high level of expression. Missense and nonsense are not common.



TP53



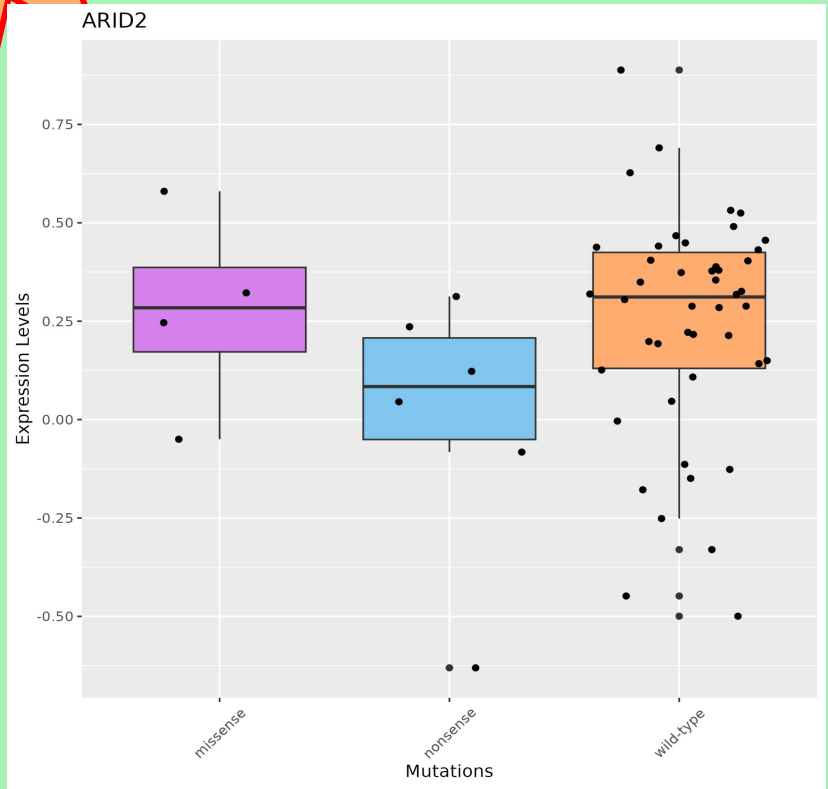
- This gene provides instructions for making a protein called “tumor protein p53”, which acts as a tumor suppressor.
- Mutations in this gene cause uncontrollable cell division and tumor formation.
- Nonsense expression levels are the lowest in this gene.

ARID2

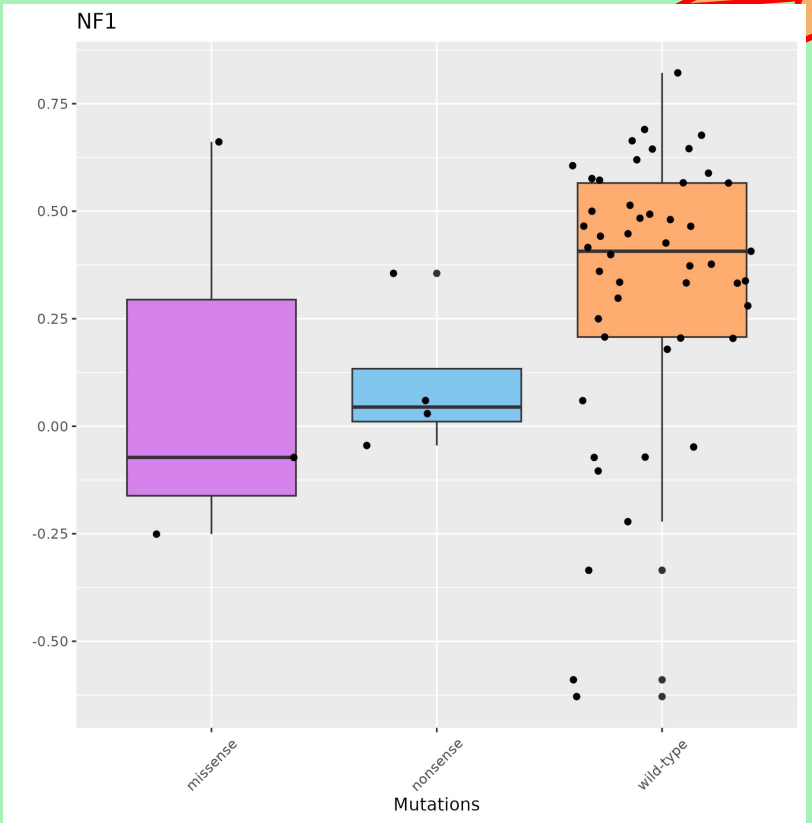
- ARID2 is a type of tumor suppressor gene. (responsible for keeping cell growth and division under control)

- When ARID2 is mutated it leads to protein loss.

- ARID2 deficiency impairs DNA repair. Meaning that if the DNA can't repair itself then the cells will become more fragile since the repair gene isn't working right.



NF1



- NF1 produces a protein responsible for cell growth and regulating it.

- A mutation of the NF1 gene leads to absence of the regulating protein and increases risk of developing benign or malignant tumors.

Presentation By: Teenage Mutant Ninja Coders

Thank you, any questions?