**Q3B891 (BRCA1)**

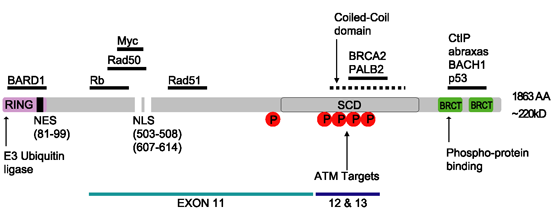
**Introduction:**

BRCA1 is a gene found in all humans and most mammals. Its primary responsibility is to provide instructions for creating a protein (Breast Cancer type 1 susceptibility protein BRCA1) that works as a tumor suppressor, which makes sure the cells are growing and dividing in a controlled way. It was mapped in 1990 and isolated from BRCA2 in 1995. It is located on the long (q) arm of the chromosome 17 at position 17q21 in humans and in most of the mammals with complete genome data available. Human DNA cannot be repaired as it should be if this gene is mutated or changed as this could stop the gene from producing vital suppressor proteins.

**Protein Structure:**

BRCA1is made of Zinc finger domain, the BRCA domain and flanked by two alpha helices (BRCT domains). Zinc ring domain is made of 40 to 60 amino acids that has 8 metal binding residues and two quartets of cysteine/histidine residues that connects to two zinc atoms. On the other hand, serene cluster domain is 1280 to 1524 amino acids long. BRCT domain approximately has 80-95 amino acids with either a single module or a multiple tandem repeat.

*Figure adapted from Creative Commons*

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**Functions:**

Human DNA can be broken due to many reasons such as natural, medical, or exposure to radiological and chemical agents. In such cases BRCA1 works as a DNA cross-linking agent to repair the broken parts by copying unbroken strands from their sister chromatids. It has been playing a vital role in copying and maintaining our DNA. It interacts with other similar proteins (BRCA2, RAD51, etc.) in order to function and repair efficiently.

**Risks of Mutations:**

A mutation to BRCA1 can cause a huge damage to our bodies. For example a mutated BRCA1 stops functioning normally, hence why it could lead to diseases like breast cancer. BRCA1 also interacts with many other proteins and regulate them to participate in embryonic development. Breast cells in women grow and divide abnormally, turn into cancer and can spread to the other parts of the body such as bones, liver, etc.

Reduction or silencing of BRCA gene can lead to breast cancer because the DNA repair mechanism uses other similar genes to compensate for the loss or silence of BRCA1. This could lead to high risk of cancer due to high chromosomal rearrangements and error-prone mechanisms of other genes. Even though BRCA1 plays a vital role in these cancers, it’s not the major cause when it comes to ovarian cancer as it is responsible for only up to 18% ovarian cancers.

A woman with a mutated BRCA1 can be susceptible to premature menopause and can become infertile younger. Having a mutated BRCA1 gene increases the risk of variety of cancers as we discussed earlier, it can also lead to breast and prostate cancers in men although it’s not frequent.

A mutated BRCA1 can lead to damaged or altered apoptosis which could lead to the disturbance of programmed cell death. Later it was discovered that the BRCA was localized within the nucleus thus all its cellular process is happening within the nucleus.

**Interactions:**

BRCA1 has many interaction partners. It regulates many other proteins to work in repairing the DNA.

* BRCA1 interacts with RAD51 to fix the damages to the DNA
* BCA1 is phosphorylated and activates the transcription of kinase
* BRCA1 and BRCA1 and a few other proteins are usually connected and make rather complex structures of proteins to work efficiently.
* It also uses PALB2 as a bridge to interact with BRCA2.

**Conclusion:**

BRCA1 is not only an important gene in repairing damaged DNA but also plays a vital role day to day cell cycle. A mutated or error-prone BRCA1 can further damage the division of the genetic material, which leads to uncontrolled growth and division of cells within human body. We cannot further clarify how it can be controlled completely as we need to put more effort and resources in to it.

**References:**

<https://varsome.com/gene/BRCA1>

<https://en.wikipedia.org/wiki/BRCA1>

<https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet>

<https://cancer.sanger.ac.uk/cosmic/gene/analysis?ln=BRCA1#overview>

<http://csbj.org/articles/e201204005.pdf>

<https://www.uniprot.org/uniprot/?query=BRCA1&sort=score>

<https://www.news-medical.net/health/BRCA1-Interactions.aspx>

<http://www.nature.com/articles/ng0701_266>

<https://breast-cancer-research.biomedcentral.com/track/pdf/10.1186/bcr557>

<https://onlinelibrary.wiley.com/doi/pdf/10.1111/j.1349-7006.2004.tb02195.x>

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4009870/>