

DR. MARTYNIUK'S RESEARCH



Background Info

X-chromosome deletion disorders like Xq27-Xq28 negatively impact the cells' mitochondria. When a cell's energy factories don't work properly it can lead to oxidative stress, which is a harmful condition where toxic molecules damage cells.

Antioxidants are substances that protect cells from oxidative damage. Researchers are testing various antioxidants to see if they can improve mitochondrial function in compromised cells, like in X-chromosome deletion patients.



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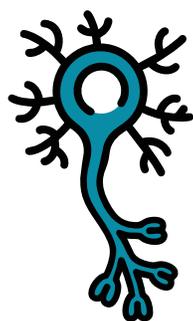
Aim 1: Test Antioxidants in Cells

Method:

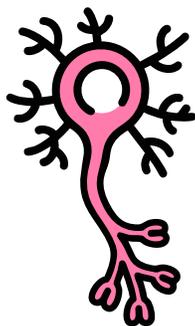
- Compare healthy cells and X-deletion cells by using tools like Seahorse Analyzers and ROS indicators to measure energy production, mitochondrial health, oxidative stress, and calcium signaling.
- Assess cellular responses to antioxidants docosahexaenoic acid (DHA) and N-acetyl cysteine (NAC).

NORMAL NEURONAL CELL

X-DELETION CELL



VS



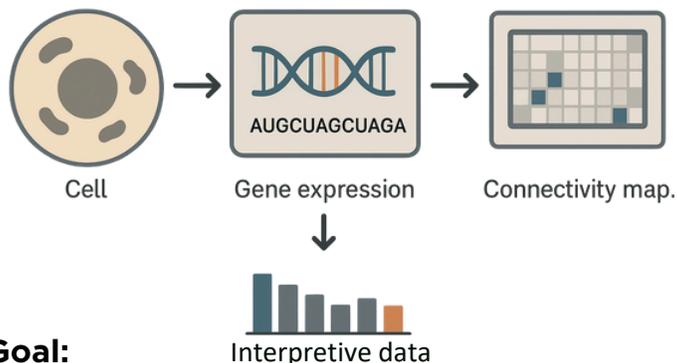
Goal:

Determine whether DHA and NAC improve mitochondrial function and reduce oxidative stress in X-deletion cells.

Aim 2: Identify new therapeutic targets

Method:

- Use RNA sequencing to identify abnormal gene expression activity in X-deletion cells compared to healthy cells.
- Use Connectivity Map to find compounds known to correct those differences in expression. This is a reverse-engineering computational approach to identifying potential therapeutics for diseases.



Goal:

Identify potential therapeutic drugs that can correct gene expression in X-deletion neurons, thus improving mitochondrial function in those cells.