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The Implications of a Neurodegenerative Disease from a Personal Perspective

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Staff Piece

How many of you play a contact sport? How many of you have friends or family members who played contact sports? Have you or anyone you know sustained a concussion in their lifetime?

These are three simple questions with much larger implications. [According to UPMC Sports Medicine](#),

- Between 1.7 and 3 million sports—and recreation—related concussions happen each year.
- Around 300,000 are football-related.
- 50% of concussions go unreported or undetected.
- 20% of high-school athletes who play contact sports, including soccer and lacrosse, will suffer a concussion this year.

I'm sure many of you at this point are wondering, What's the big deal? Why should I care?

When I was six-years-old, my uncle passed away from [Amyotrophic Lateral Sclerosis \(ALS\)](#). ALS is a neurodegenerative disease that affects the neurons in the spinal cord and brain. The muscles begin to lack nourishment, and become so weak that they degrade. The spinal cord will begin to develop sclerosis, the hardening of the neurons and muscles. This eventually results in nerves not being able to fire, muscles becoming fragile, and atrophy.

My uncle, [Jim](#), was only 43 when he passed. Six years of his life was spent battling this disease. This means that since I was born, he was living with this diagnosis. My memories of a healthy Jim are essentially nonexistent. He eventually realized something was wrong when he woke up one day unable to tie his shoes. He was confused, not understanding why a part of his daily routine was simply impossible. After many trips to the doctor, and many different tests, the final diagnosis of ALS shook him, as well as our entire family.

Jim, by the way, was not unhealthy by any means. He was a star quarterback and safety on the football team throughout high school, even winning a championship in 1979. He made the All-League Team his junior and senior year, All State Third Team defensive back as a junior, All State Team quarterback as a senior, and was selected to his alma mater's Athletic Hall of Fame.

He continued to play football at Lehigh University, earning his letter three of his four years there as a free safety, ranking sixth in the country in interceptions. Jim was bright and talented: gifted, if you will.

Jim loved the game so much, nothing could stop him. He had sustained multiple concussions throughout his lifetime, even playing with some or refusing to see trainers so that he could continue to play in that week's game.

When he died thirteen years ago, little was known about ALS or potentially connected illnesses. ALS is now known to be closely linked to a disease known as [Chronic Traumatic Encephalopathy \(CTE\)](#). CTE occurs when proteins called "tau" proteins conglomerate in the brain, causing neuronal blockage and the death of brain cells. What causes CTE, you might ask? Repetitive Head Impacts (RHI)—most commonly known as concussions. CTE is commonly found in athletes and military veterans due to a heightened rate of RHI in both groups.

Sadly, there is no cure for CTE—or ALS for that matter—at this point. In fact, CTE cannot even be diagnosed during the patient's lifetime yet. Due to its location in the brain, diagnosis can only occur *after* death. A thin slice of brain tissue from the deceased patient will be stained and analyzed under a microscope to look for tau conglomerations and characteristic patterns of those proteins within the brain.

Athletes and veterans who have died of ALS—which [can be diagnosed](#) during the patient's lifetime thanks to EMGs, MRIs, blood tests, and spinal taps—have had brain tissue analyzed postmortem. It gave a shocking result when a number of those people who passed from ALS had those characteristic tau protein conglomeration patterns within the brain, indicating they *also* suffered from CTE. The correlation of CTE and ALS remains strong, namely in collegiate/professional athletes, military veterans, and car accident victims.

We are playing a sad game of chicken or the egg in science right now. Which comes first: ALS or CTE? How are they affecting each other? What can we as a community do to spark conversation and inform people about the serious implications of these diseases?

That is why I am sitting here typing away on my laptop, fighting to hold back tears. As a scientist, niece, and friend, I am here to start that conversation. Talk to your teammates about the severity of concussions, ask your coaches and athletic trainers to provide more information, inform your friends, talk to your parents, and read journal articles.

It seems bleak and obvious that concussions are bad, and neurodegenerative diseases are in no one's life plan. What people don't talk about is how hard it is to watch a loved one live their life from a wheelchair with a feeding tube, unable to speak or care for themselves. Had my family known about potential links between CTE and ALS when Jim passed, we would have had his brain donated for research. Unfortunately, we are living with too many "What ifs?" to go back in time and fix them.

So, I urge you to start advocating for yourself, your friends, your family members, and even your favorite sports players. Fight for those people who can no longer fight for themselves, and support the scientists who go to work every day with the intentions to find the answers people desperately need.

For more information on ALS and CTE related research:

1. <http://www.bu.edu/cte/>
2. http://web.alsa.org/site/PageNavigator/blog_011718.html
3. <https://academic.oup.com/jnen/article/77/12/1091/5123933>

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2. <http://www.alsa.org/about-als/what-is-als.html>
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4. <https://concussionfoundation.org/CTE-resources/what-is-CTE>
5. <https://www.mayoclinic.org/diseases-conditions/amyotrophic-lateral-sclerosis/diagnosis-treatment/drc-20354027>

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