# Readily-available drugs may reduce devastating symptoms of Tay-Sachs disease

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Biology professor Suleiman Igdoura, McMaster University, and his team have found FDA-approved drugs may help ease symtpoms of Tay-Sachs disease. Credit: McMaster University

A team of researchers has made a significant discovery which may have a dramatic impact on children stricken with Tay-Sachs disease, a degenerative and fatal neurological condition that often strikes in the early months of life.

Available drugs may dramatically ease a child's suffering, say scientists.

"There is hope for this disease," says Suleiman Igdoura, lead researcher of the study and an associate professor of biology at McMaster University. "Imagine what that could mean for parents who have a child diagnosed with this incurable condition, who may have only a few years with their child."

Tay-Sachs is a genetic disorder caused by the absence of vital enzymes which are involved in the breakdown of waste within cells. Without those enzymes, waste accumulates and eventually destroys healthy cells, leading to blindness, paralysis, mental retardation and eventually death.

Igdoura and his team have found that when a key protein in the brain—known as TNFa—is removed, some of the devastating symptoms of Tay-Sachs and its close relative Sandhoff, were much less severe when tested in mice. Those symptoms include spasms, muscle wasting and loss of neurological function.

The findings are significant because the protein can be managed by FDA-approved drugs, readily available on the market.

"With Tay-Sachs and Sandhoff, we have very little to offer families in terms of therapeutics to help their children," says Igdoura.

"These are orphan diseases where there are not many medications available. But we feel this is a significant step in improving quality of life and quite possibly extending lives."

Children who are diagnosed with Tay- Sachs early in life, typically die before they reach the age of four or five.

"There are distinct stages within the disease, so we wanted to find targets

we could interfere with, to delay the terrible outcome or halt it altogether," explains Igdoura.

Using mice which were genetically altered to mimic Tay-Sachs and Sandhoff, researchers found levels of TNFa rose significantly during the early stages of the diseases.

But when TNFa was subsequently removed, there was a significant improvement in the lifespan of the mice and neurological function.

"We also found that neurons didn't die as early as they do with the disease, so we delayed the progression as well," says Igdoura. "We have identified a molecule that is the culprit and we believe there are drugs available to stop it."

The research is published online in the journal *Human Molecular Genetics*.

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