



Miglustat for treatment of Niemann-Pick C disease a randomized controlled study

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Plain Language Summary: Results of a randomized, controlled study of miglustat for treatment of NPC

What is this study about?

This research study tested whether a medicine called miglustat can help people with Niemann-Pick Disease Type C (NPC). NPC is a rare genetic disease that affects how the body handles certain fats, causing serious problems in the brain, liver, spleen, and other organs over time.

What did the researchers do?

The scientists conducted a randomized controlled trial, which is considered the gold standard for testing medicines. This means they randomly divided patients into two groups - some got miglustat and others got a placebo (fake medicine). Neither the patients nor the doctors knew who was getting which treatment until the study ended. This type of study design helps ensure the results are reliable and not influenced by bias.

- **Who Participated:** The study included 29 patients aged 12 years or older who had NPC. The study was conducted over 12 months.
- **How They Were Divided:** These patients were randomly put into two groups:
 - 20 patients received miglustat (200 mg, three times a day).
 - 9 patients received "standard care," which means they got the usual treatment without miglustat.

How does miglustat work?

Miglustat influences Niemann-Pick disease type C (NPC) in several ways. One mechanism of action involves blocking the body's production of specific fatty substances called glycosphingolipids. This particular approach is known as substrate reduction therapy. Additionally, it's thought that miglustat has other, more intricate effects that are not yet fully understood.

What this means for patients:

NPC typically gets worse over time, affecting how people move, think, and function. Finding a medicine that could slow down or stop these problems would be life-changing for patients and their families. The research showed that the medicine can stabilize or slow down specific NPC symptoms. Later studies have even suggested that miglustat may help people with NPC live longer. This study paved the way for miglustat to become an important treatment option for NPC patients worldwide, giving hope to families dealing with this challenging disease.

While miglustat has been approved for NPC patients in many countries, its use in the U.S. for NPC remains off-label (meaning the FDA didn't formally approve it for NPC). The FDA approved Miplyffa to be used in combination with miglustat for NPC.