



Plain Language Summary

Final report of the NPC Registry on the treatment outcomes following continuous miglustat therapy in patients with NPC

Citation: Patterson MC, Vecchio D, Jacklin E, et al. Treatment outcomes following continuous miglustat therapy in patients with Niemann-Pick disease Type C: a final report of the NPC Registry. *Orphanet J Rare Dis.* 2020 Mar 16;15(1):70. doi: 10.1186/s13023-020-1346-x. PMID: 32178700; PMCID: PMC7075000.

<https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-01363-2>

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What was this study about? This study looked at the long-term effects of a medicine called miglustat in people with Niemann-Pick Disease Type C (NPC). NPC is a rare, serious genetic disease where the body can't properly move fats, leading to a buildup of these fats in cells, especially in the brain, liver, and spleen. This buildup causes a range of problems, including issues with movement, speech, and thinking. The study aimed to understand how well miglustat works over many years to slow down the disease's progression and how safe it is with continuous use.

Who participated in the study? The study included 120 patients with NPC who were receiving miglustat as part of their regular care. These patients were of various ages, from children to adults, and their information was collected through the International Niemann Pick Disease registry specifically for NPC patients.

What was tested? The researchers evaluated miglustat, which is a medicine approved for treating NPC. Miglustat works by reducing the amount of certain fatty materials that build up in the cells of people with NPC. The goal was to see if taking miglustat continuously could slow down the worsening of NPC symptoms over time.

How was the study done? This was an observational study that used data from the Niemann-Pick Disease Type C Registry. This means that instead of a controlled trial where patients are assigned to different groups, researchers collected real-world information from patients who were already taking miglustat as part of their standard treatment. Patients were followed for up to 10 years, and their symptoms, disease progression, and any side effects were recorded.

What were the main findings? The study found that:

- **Slower Disease Worsening:** Patients who continuously took miglustat showed a slower worsening of their neurological symptoms (like problems with walking, speech, swallowing, and eye movements) compared to what is typically seen in NPC patients who are not treated.
- **Long-term Benefits:** The positive effects of miglustat in slowing down the disease seemed to continue over many years of treatment.
- **Safety:** Miglustat was generally well-tolerated over the long term. The most common side effects were related to the stomach and gut, such as diarrhea or weight loss, but these often improved over time or could be managed.
- **Early Treatment:** The study suggested that patients who started miglustat earlier in their disease course, especially younger patients, tended to have better long-term outcomes.

What do these findings mean? The results of this study provide strong evidence that continuous miglustat therapy is a valuable long-term treatment for Niemann-Pick disease type C. It helps to slow down the progression of neurological symptoms, which are a major challenge for patients. The findings also highlight the importance of starting treatment early to achieve the best possible outcomes for individuals living with NPC.

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.