

Treatments for LoF (Loss of Function):

1. L-Serine: This is an amino acid (the building blocks of protein) found in many foods such as corn, soy, chicken, turkey, eggs and almonds. In cells, the active form of L-Serine is D-Serine. At the cellular level in the laboratory, D-serine has been shown to help neurons with LoF mutations to function better. D-Serine is not safe to be given directly, so the L- form is used which is then changed to the more active D- form inside the body.

L-Serine has been studied in detail in a clinical case, but more studies are being carried out to confirm these findings. Only few adverse effects have been described, and it appears to be, overall, well tolerated. However, the erroneous administration to a GoF child was associated with severe epileptic episodes.

The medical team led by Dr. García-Cazorla from the Hospital Sant Joan de Deu, in Barcelona, started in 2020 with the first clinical trial in the world for the food supplement L-Serine, to determine the tolerability and efficacy of this treatment in GRIN children with loss of function (LoF). This study was replicated by several hospitals around Europe. The results of the Spanish clinical trial are expected to be published towards the summer of 2023.

Here are the links for the different publications about L-Serine

L-Serine Clinical Case:

https://www.science.org/doi/10.1126/scisignal.aaw0936

https://link.springer.com/article/10.1007/s13311-021-01173-9

L-Serine Clinical Trial:

https://www.clinicaltrials.gov/ct2/show/NCT04646447? term=Serine&cond=GRIN&draw=2&rank=1

L-Serine Case Reports

https://pubmed.ncbi.nlm.nih.gov/34997442/

2. Piracetam (Nootropil): This is a neurotropic medication that improves the metabolism of neurons. It is available as an over-the-counter supplement in some European countries but in others it is only available with a prescription. Piracetam also affects the NMDA neuroreceptor. The use in GRIN is still anecdotal.

https://www.ptfarm.pl/pub/File/Acta_Poloniae/2005/5/405.pdf

If you want to learn more about the Clinical Trials for GRIN disorders please check our Clinical Trial section under the RESEARCH tab.