SPG76 Natural History Study

An invitation to the neurologist of a patient with SPG76

Dear Doctor,

Your patient has given you this note because they would like you to consider contributing to an international natural history study of SPG76, the spastic paraplegia caused by biallelic CAPN1 variants.

About the study

The study is led by Dr. Ziv Gan-Or at The Neuro, McGill University, Montreal. We are building a combined natural history dataset from all published and unpublished SPG76 cases, so the field can describe how the disease progresses - for example, the age at which patients first need a walking aid or a wheelchair - and whether genotype affects the course. No single center has enough patients to answer these questions, and they are what future treatment trials will need.

What we are asking

1. Share de-identified clinical information on your patient - the CAPN1 variants, age at onset, the main clinical findings, and any scale scores or tests already done (for example SPRS, nerve conduction studies or brain MRI).
2. Add follow-up information over time, if you have it - especially the age at first use of a walking aid and the age at wheelchair dependence.
3. Add any other SPG76 patients you follow, published or not.

Only de-identified clinical information is needed - no names, dates of birth or other identifiers, and no samples. It draws on information from your routine care, so no extra visits or tests are needed. The data comes to our team at The Neuro, McGill University, and can be shared in whatever way your institution requires. Contributing clinicians will be invited to be co-authors on the resulting publication.

How to take part

Please email Dr. Ziv Gan-Or at **ziv.gan-or@mcgill.ca** with "SPG76 natural history" in the subject line. We will send you a short data sheet and take it from there.

About the SPG76 Research Foundation

The SPG76 Research Foundation joins us in this work. It is a patient-founded nonprofit, started by Omar Alnuaimi, who lives with SPG76. The foundation connects patients, clinicians and researchers; it does not collect or hold any patient data, so please send information directly to the study team.

Thank you for considering it.

With best regards,

Dr. Ziv Gan-Or - The Neuro, McGill University, Montreal, Canada
Omar Alnuaimi - SPG76 Research Foundation (spg76research.org)