

Cutaneous Neurofibromas

Cutaneous Neurofibromas Group

Cutaneous neurofibromas (cNF) are benign peripheral nerve sheath tumors and a hallmark feature of NF1. While cNFs do not undergo malignant transformation, disfigurement and associated pruritus may significantly impact quality of life. There is no effective medical therapy for cNFs, and the current management is limited to surgical removal of the lesions amenable to surgery. Substantial progress has been made in the understanding of the biology of neurofibromas, which may allow for the acceleration of the development of effective therapies. Goals of the cNF working group will include the identification of research priorities for cNFs and the development of recommendations for endpoints, designs, and outcome measures for clinical trials directed at cNFs.

Current group Leaders: Ashley Cannon, PhD, MS, CGC & Kavita Sarin, MD

Current Project

As a part of the REINS Cutaneous Neurofibroma Group initiative, we are conducting a Delphi about the outcome domains to be measured in the upcoming clinical trials on cutaneous neurofibromas (cNFs).

The aim of this current project is to develop a core outcome domain set which will define the minimum set of outcomes to be measured in the coming clinical trials for cNFs. This will help avoid from the very beginning lack of uniformity in the outcomes reported and will eventually allow to compare treatment effectiveness across trials.

A candidate list of outcome domains has been generated through a systematic review of the literature, focus groups with NF1 patients and patient representatives, focus groups with international NF1 experts, and other healthcare professionals involved in the care of NF1 patients. The purpose of this eDelphi is to reach a consensus on the most important outcome domains to be reported in clinical trials on cNFs.

To reach consensus, we are conducting a Delphi survey that will consist of two online surveys, followed by a consensus meeting (hybrid meeting), and a third final online survey.

The first survey will be launched in February 2022 and second survey is planned in March 2022. The consensus meeting will take place during the CTF NF meeting in June 2022, and the third survey will be launched in July 2022. Participants will have a 2 weeks window to complete the survey after launch.

If you are interested in participating in the Delphi, feel free to send an email to christina.bergqvist@aphp.fr before January 15.

Publications

- 1.) Ashley Cannon, Dominique C. Pichard, Pamela L. Wolters, Sarah Adsit, Gregg Erickson, Andrés J. Lessing, Peng Li, Whitney Narmore, Claas Röhl, Ten Rosser, Brigitte C. Widemann, Jaishri O. Blakeley, Scott R. Plotkin. Perspectives of Adults with Neurofibromatosis 1 and Cutaneous Neurofibromas. *Neurology* 2021. Aug 2021; 97 (7 Supplement 1) S15-S24; DOI:10.1212/WNL.0000000000012425. [\[PDF\]](#)
- 2.) Sheilagh Maguiness, Yemima Berman, Nathan Rubin, Melissa Dodds, Scott R. Plotkin, Claire Wong, Christopher Moertel. Measuring the Effect of Cutaneous Neurofibromas on Quality of Life in Neurofibromatosis Type 1. *Neurology* 2021. Aug 2021; 97 (7 Supplement 1) S25-S31; DOI:10.1212/WNL.0000000000012427. [\[PDF\]](#)
- 3.) Raquel D. Thalheimer, Vanessa L. Merker, K. Ina Ly, Amanda Champlain, Jennifer Sawaya, Naomi L. Askenazi, Hamilton P. Herr, Jennifer L.W. Da, Just in T. Jordan, Alona Muzikansky, Elizabeth Morehouse Pearce, Fernanda H. Sakamoto, Jaishri O. Blakeley, R. Rox Anderson, Scott R. Plotkin. Validating Techniques for Measurement of Cutaneous Neurofibromas. *Neurology* 2021. Aug 2021; 97 (7 Supplement 1) S32-S41; DOI:10.1212/WNL.0000000000012428. [\[PDF\]](#)
- 4.) Deeann Wallis, Anat Stemmer-Rachamimov, Sarah Adsit, Bruce Korf, Dominique Pichard, Jaishri Blakeley, Kavita Y. Sarin. Status and Recommendations for Incorporating Biomarkers for Cutaneous Neurofibromas Into Clinical Research. *Neurology* 2021. Aug 2021; 97 (7 Supplement 1) S42-S49; DOI:10.1212/WNL.0000000000012426. [\[PDF\]](#)

Presentations

Resources (click to download)

[Adult cNF Survey: Patient Attitude and Beliefs Regarding Cutaneous Neurofibromas and Treatment](#)

- This survey is used to assess the perspective of adults with Neurofibromatosis Type 1 regarding cutaneous neurofibromas for designing clinical trials. The results of this anonymous survey will be published in an upcoming *Neurology* supplement of REINS manuscripts.