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Background: Cutaneous neurofibromas (cNF) affect greater than 99% of patients with neurofibromatosis type I and negatively affect patients' quality of life. cNF usually present around puberty and increase in both number and size with age. Current cNF standard of care is to wait until patients can no longer tolerate the associated psychosocial and physical burdens and then surgically excise lesions. Removing all lesions is usually infeasible due to the large number of cNF and postsurgical scarring. Our objective is to detect nascent cNF using non-invasive optical imaging methods and enable their early treatment to halt cNF's development into visible skin tumors.

Study Design/Materials and Method: We imaged nine skin areas on three subjects with cNF using a color camera, spatial frequency domain imaging (SFDI) and optical coherence tomography (OCT). SFDI quantified the scattering and absorption properties of human skin over a wide field-of-view and provided maps of possible nascent cNF due to different optical properties between cNF and surrounding skin. High-resolution, three-dimensional OCT images were recorded to study the skin's microstructure and microvasculature changes associated with cNF.

Results: cNF visible in camera images were also visible in SFDI scattering coefficient maps and characterized by a lower scattering coefficient than surrounding tissue. Barely perceptible cNF in camera images were clearly visible in SFDI scattering maps. Suspect cNF lesions were found in the scattering map but not in camera images. OCT images confirmed that light scattering is reduced inside all perceptible or barely perceptible cNF. Both abnormal tissue structure and microvasculature were observed in the suspect nascent cNF areas.

Conclusion: Optical scattering in cNF may be a viable cNF marker in skin and SFDI is capable to detect cNF lesions including nascent cNF which can be analyzed at higher resolution with OCT to study tumor development and response to candidate therapies.