

Self-evaluation of disease visibility in patients with neurofibromatosis type 1: development of an online scale

Autoavaliação da visibilidade da doença em pacientes com neurofibromatose tipo 1: desenvolvimento de uma escala on-line

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Dear Editor,

Neurofibromatosis type 1 (NF1) is a rare genetic disorder that affects approximately one in every 3,000 individuals, regardless of sex or ethnic background.^{1,2} The disorder is caused by a mutation in the NF1 gene, which functions as a tumor suppressor.² The phenotypic expression of NF1 is highly variable. Nearly all patients develop small benign skin tumors that increase in number and visibility with age. In addition, 30% of adults with NF1 have visible plexiform neurofibromas.³

The NF1's visibility of NF1 manifestations is strongly associated with patients' quality of life and well-being of individuals.¹⁻⁴ Research suggests that patients' self-perception of their appearance is more important to them than visibility rated by external ratings.^{1,4} Therefore, the assessment of disease visibility in NF1 should ideally be based on self-reported questionnaires rather than ratings by specialists.⁴

The aim of this study was to develop a self-rating scale, based on the Ablon Visibility Scale5 to measure NF1 visibility scale5 (Table 1). The scale is designed for remote access and self-administration by patients. It consists of eight yes/no ques-

Letters

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tions, with NF1 visibility is classified into three levels: 1 (mild), 2 (moderate), and 3 (severe), based on the combination of responses (Table 2). To ensure clarity, we used simple language to make technical terms easily understandable to patients. For remote use, the scale was implemented via Google Forms, incorporating images from the Dermatology Information System to assist individuals in completing the assessment (<https://doi.org/10.6084/m9.figshare.14442107.v4>).

To assess the feasibility of the scale, we conducted an observational clinical study involving seven adult patients with NF1. Six of these patients had previously participated in research

conducted by our group in 2014, during which they were classified in person using Ablon Scale.² Between April and May 2021, these individuals used the newly developed NF1 visibility self-evaluation scale to rate themselves and were subsequently rated remotely by a dermatologist using the same tool. Table 3 compares the 2014 face-to-face classifications with both the self-assessments and the dermatologist's remote evaluations from 2021. Four patients rated their disease as more visible than in the 2014 face-to-face assessment, but their self-ratings matched the dermatologist's remote classification in 2021. This discrepancy may be attributed to the natural progression of the

TABLE 1: Ablon Visibility Scale of neurofibromatosis type 1

Degree 1 – Mild case	Tumors are generally not visible outside of normal clothing areas. Gait and posture appear normal to casual observation, although there may be a significant number of neurofibromas under clothing and minor skeletal symptoms.
Degree 2 – Moderate case	Some tumors are visible on the neck, face, and hands. Mild scoliosis or other skeletal features may be present, but there is no noticeable limp.
Degree 3 – Severe case	There are many tumors on the face. An optic glioma (tumor) may affect vision and the eye socket. Severe scoliosis or skeletal features are present, resulting in a noticeable limp.

TABLE 2: Items for self-evaluation scale of neurofibromatosis type 1 visibility

Feature	Degree of visibility
I have café-au-lait spots (brown patches) on my body	Can be degree I, II, or III*
I have small neurofibromas (pellets on the skin) around my body that are not visible (they are covered by clothes)	Degree I
I have some neurofibromas (pellets on the skin) around my body that are apparent on my neck, face, and hands	Degree II
I have many neurofibromas (pellets on the skin) around my body that are apparent on my neck, face, and hands	Degree III
I have mild scoliosis (deviation in the spine), which is not perceived (noticed) by other people	Degree II
I have more severe scoliosis (deviation in the spine), which is perceived (noticed) by other people	Degree III
I had to have surgery because of scoliosis (deviation in the spine)	Degree III
I have asymmetry of the face, that is, there is a difference between the two sides of my face	Degree III
I have other health problems because of neurofibromatosis type 1. If so, please describe them	—

* Consider degree I if marked separately.

TABLE 3: Comparison of classifications by Ablon Visibility Scale (2014) and the online NF1 visibility self-evaluation scale (2021)

Patient	Ablon Visibility Scale (face-to-face evaluation – 2014)	Online NF1 visibility self-evaluation scale	
		Self-evaluation – 2021	Evaluation made by the researcher – 2021
1	Degree I	Degree I	Degree I
2	Degree II	Degree III	Degree III
3	Degree I	Degree I	Degree I
4	Degree I	Degree II	Degree II
5	Degree II	Degree III	Degree III
6	Degree I	Degree II	Degree II
7	—	Degree III	Degree III

disease. Overall, there was complete agreement between the patient self-assessments and the dermatologist's remote evaluations using the NF1 visibility self-evaluation scale.

Visibility and severity of NF1 are important to distinguish. Visibility refers to the outward appearance of a fully clothed individual and how easily symptoms are noticed during casual, impersonal interactions. Many individuals with NF1 may not have visible phenotypic features in commonly exposed areas, but they may have numerous tumors and/or café-au-lait spots in areas that could become visible in certain situations, such as on the beach or during intimate contact.^{3,5} In contrast, severity encompasses both clinical and cosmetic factors, including impact on lifestyle, mobility, and even life-threatening complications.¹⁻⁴ While visibility can be self-assessed by the patient, as it involves only external features that can be observed and reported, severity assessment requires professional analysis. This assessment considers the extent of dermatologic involvement and the presence of disabling complications. Both visibility and severity of NF1 are related to psychosocial well-being and the need for appropriate support.¹⁻⁴

The visibility scale we developed has proven to be a valuable tool that can be used in clinical research as well as in the care of individual patients. By allowing patients to apply the scale themselves, it emphasizes the importance of their personal

perception of their condition and can help improve their understanding of the disease and its progression.

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Availability of data and materials

The datasets generated and/or analyzed during the current study are available from the corresponding author upon reasonable request.

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Conflict of interest

The authors declare no conflicts of interest.

Ethics approval and consent to participate

This study was approved by the Human Research Ethics Committee at Universidade Federal de São Paulo (CAAE 29747620.0.0000.5504), and all participants provided informed consent. ●

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