



Response Evaluation In Neurofibromatosis Schwannomatosis INTERNATIONAL COLLABORATION

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REiNS Patient Reported Outcomes (PRO) Working Group

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On behalf of the REiNS PRO group



Response Evaluation In Neurofibromatosis Schwannomatosis
INTERNATIONAL COLLABORATION

Assessing Perceived Disfigurement in NF Clinical Trials



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Definitions of disfigurement

- Visible differences that can affect aesthetic, functional, or psychological well-being
- Visible difference marked by a condition or injury that affects appearance

Symptoms related to disfigurement

- NF1
 - PNs, cNFs, scoliosis, other bony issues, café au lait macules, short stature
- NF2
 - Orbital meningiomas, facial weakness, ptosis, muscle wasting
- Schwannomatosis
 - No symptoms identified
- Other visible differences across NF types:
 - Walking with cane, wheelchair, scarring



Current Goals

1. Create a rating scale to assess features of disfigurement that are (potentially) changeable in a clinical trial
2. Assess disfigurement as a primary (physical) construct (rather than more distal issues, e.g., social-emotional functioning, that are related to disfigurement)
3. Develop patient-self report (8+) and parent report forms (all ages)



Disfigurement Scales

Rate the severity of disfigurement based on your first impression.

1 2 3 4 5 6 7 8 9 10

Not
disfigured

Very
disfigured

Lyford-Pike et al., 2018



Disfigurement Scales (cont'd)

No instructions given

1	2	3	4	5	6	7	8	9
minimally visible disfigurement (visible at close range only)			moderately visible disfigurement			very visible disfigurement (visible from afar)		

Chen et al., 2017



Item 1

Disfigurement Rating Scale Patient Self-Report (ages 8 and older)

Disfigurement is a visible difference on the face or other parts of the body. Some people have disfigurement due to their NF.

The medical team has chosen your _____ as an area of disfigurement to check on for this study.

1. How disfigured is your _____ **today?**

0 1 2 3 4 5 6 7 8 9 10
Not at all Very
disfigured disfigured



Item 2

2. Is your _____ the most important disfigurement on your face or body?

Yes No

If yes – end of questions.

If no, please answer questions 3 and 4.

3. Where is your most important area of disfigurement on your face or body?

4. How disfigured is that area **today**?

0 1 2 3 4 5 6 7 8 9 10

Not at all
disfigured

Very
disfigured



Future Directions

- Administer to patient volunteers including children to get input on wording, instructions, etc.
- Share updated version with patient representative working group
- Review scales on related domains – body image, appearance-related self-esteem, satisfaction with appearance
- Explore possibility of qualitative research study to validate tools (funded by PCORI, CTF?)



Questions for the group

1. How feasible would it be to add a figure of a person and circle the area of disfigurement?
2. How much would these items need to be modified in accordance with different clinical trial targets (cNF, bony changes, facial weakness)?
3. What is the best way to validate these items (ongoing or upcoming trials, separate study)?



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